Sexual Precocity

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Sexual Precocity

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To
Geraldine

Foreword

ALTHOUGH NUMEROUS PAPERS ON SEXUAL PRECOCITY HAVE APPEARED in the literature most of these consist of reports on only one or several cases In many instances modern methods of endocrinologic study were not made Dr Jolly has performed a valuable service in collecting and studying intensively 69 patients with this disorder throughout England His monograph presents a wealth of factual data of importance to the endocrinologist and the student interested in the subject It fulfills a real need by shedding additional light on the relative frequency of different types of sexual precocity and on the methods of differential diagnosis In addition the psychologic aspects of the disorder are given careful consideration

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H J

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Sexual Precocity

ONE

Introduction

FROM THE VERY EARLIEST DAYS OF MEDICINE INSTANCES OF SEXUAL PRECOCITY have attracted the interest of doctors, and the first on record is that of Mandeslo (quoted by Lenz 1913) who in 1658 reported a girl whose menstruation had commenced at three years of age and who gave birth to a son when six years old Possibly the best known of these older examples is that of Anna Mummenthaler first reported by Albrecht Haller in 1751 (quoted by Ahlfeld 1898) In this girl menstruation first occurred at two years she experienced a stillbirth at nine the menopause at 52 and died at 75 years of age

In 1902 Williams was able to collect from the literature of the 19th century, 105 examples of precocious sexual development but interesting as are these older records from an historical point of view they add little to our knowledge of the aetiology In fact as Novak (1944) points out even as recently as the papers of Lenz (1913) Reuben and Manning (1922 and 1923) and Neurath (1928), where many examples from the literature have been collected together, this same problem of aetiology arises and cannot be solved from the point of view of modern pathology

This study was awarded the Raymond Horton Smith Prize in the University of Cambridge for 1950–51

and Evans (1930) at least a third of the pulmonary circulation must be occluded for paradoxical embolism to take place

In cases of patent ductus arteriosus paradoxical embolism is shown by the occurrence of infarcts in the pulmonary and systemic circulations when infective endocarditis is present The presence of vegetations at the aortic orifice of the ductus permits infective particles to be carried along the ductus from left to right with resultant infarcts in the lungs Certain authors have correctly considered this type of paradoxical embolism to be more frequent than that which occurs with the auricular septal defect

Finally paradoxical embolism in cases where a dextroposed aorta lies astride an interventricular septal defect may be the cause of unexpected death after surgical operations in infected cases Farre (1814) appears to have been the first to record a cerebral abscess in cyanotic congenital heart disease His case was a cyanotic boy of nine and there was no bacterial endocarditis Peacock (1866) was of the opinion that most congenital cyanotic cases died of some cerebral incident Ballet (1880) observed that death from cerebral abscess was frequent in cyanotic congenital heart cases Abbott Lewis and Beattie (1923) collected six cases of the tetralogy including two of their own where death was the result of a cerebral abscess due to paradoxical embolism They emphasize the frequency of thrombosis in the right chambers and peripheral veins and the direct path afforded by the overriding dextroposed aorta Wechsler and Kaplan (1940) summarized fourteen cases of cerebral abscess in congenital heart disease and in ten the abscess was on the right side Gates Rogers and Edwards (1947) state that there are thirty reported cases

Examination of the literature of cerebral abscess associated with cyanotic congenital disease shows that its occurrence is most frequent where there is a venous arterial shunt allowing re circulation of venous blood in the systemic circulation without its passage through the lungs Filtration of organisms by the pulmonary capillaries is thus avoided Important anatomical defects in this respect are the tetralogy of Fallot Eisenmenger complex cor triloculare biatriatum and the atrial septal defect in failure The source of infection is most often sepsis in the throat or ear tonsillectomy or dental extraction Bacterial endocarditis is almost invariably absent Cerebral abscess can occur at any age but rarely below one year The oldest reported case was 57 years Attempts at surgical cure have been made and operation was successful in the case of Smolik Blattner and Heys (1946)

It is almost impossible to be completely certain of the diagnosis of paradoxical embolism in cases where an auricular septal defect is a latent abnormality although the condition may be suspected in cases which show infarctions in both circulations It is reasonable to be more positive in diagnosis when embolism occurs in both circulations in

cases of patent ductus arteriosus and the tetralogy of Fallot The occurrence of a brain abscess in the latter condition in the absence of local predetermining causes is almost certainly due to paradoxical embolism However the diagnosis will rarely be positively made during life although it may be suspected if the clinical phenomena are kept in mind *Usually the extreme rapidity of death following upon important* pulmonary embolism gives small opportunity for the recognition of all the components of the condition The essential picture from a diagnostic point of view is one in which pulmonary embolism is followed by numerous emboli in the systemic circulation The diagnosis is naturally strengthened by the presence of congenital heart disease The prognosis is in general exceedingly poor

CHAPTER II

EMBRYOLOGY AND MORPHOLOGY

Some knowledge of the salient facts of embryology and morphology are essential for a proper comprehension of congenital heart disease The deformed human heart due to arrested development at an early stage may broadly resemble the heart of one of the more primitive vertebrates This reversion towards a more primitive morphological type was first noticed by Meckel (1812) and has been commented upon by numerous observers since that time Actually the critical period in the development of the human heart is before the septa have closed that is in the fifth to eighth week of embryonic life At this time the complex processes of torsion involution of the bulbus and formation of the septa are taking place Any interruption in or arrest of these processes may result in grave anatomical abnormality and call for remarkable readjustment of the organism to meet changed and disabling conditions

The fundamental purpose controlling the development of the heart is the formation of two circulations the pulmonary and systemic in such a way that they are in communication and an exchange of blood may be effected It is thus necessary for the processes of development to effect a crossing of the circulation so that the pulmonary vein enters the aortic side of the heart and the pulmonary artery may lie in the path of blood from the vena cavae The actual mechanical stimulus causing development to proceed along certain well defined lines remains a matter for debate By some it is held to be an inherent capacity possessed by the primitive cardiac cells by others it is thought to be linked with haemodynamic processes Space precludes any detailed discussion of these and other embryological factors and only a brief outline will be given of those developmental processes which are of help in the interpretation of cardiac defects

EARLY STAGES In its earliest stages the human heart bears some resemblance to that of the primitive fishes It is a simple tube like structure consisting successively of a truncus arteriosus bulbus cordis common ventricle common auricle and sinus venosus This tubular structure is anchored at its ends by the vessels entering the truncus and the sinus venosus Such an anatomical state exists at the end of the fourth week In the fifth week with the relatively rapid growth and expansion of its component parts the tube becomes kinked upon itself and rotated upon its long axis It assumes an S shape the two

curvatures of which are not in the same plane the lower curve being to the left and behind (fig 1) The common ventricle is anterior with the bulbus passing upwards and to the left the loop so formed being termed the bulboventricular loop the bulboventricular cleft intervening between these two structures The auricle and sinus venosus are situated slightly above and posteriorly In the fifth week there also appears the first signs of the septum primum the bulbar ridges and the endocardial cushions of the atrial canal As the auricle develops it expands laterally

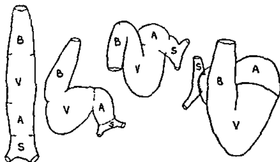


FIG 1 The early stages of the development of the heart (After Pichon)
A auricle B bulbus S sinus venosus V ventricle

and tends to overlap the bulbus which embeds itself in the auricular furrow The sinus venosus opens into the right side of the auricle and is still a separate sinus into which enter the venae cavae The atrial canal connecting the common auricle and ventricle opens into the left side of the latter In the sixth week the septum primum fuses with the endocardial cushions of the atrial canal and the septum primum is formed the septum secundum appears the truncus becomes completely divided and the endocardial cushions destined to form the valves are present The seventh week is marked by the disappearance of the sinus venosus into the auricle the formation of the coronary sinus from the duct of Cuvier and the development of the septum spurium and foramen ovale In the eighth week the heart assumes its familiar configuration and the interventricular septum closes In the subsequent weeks until term further differentiation of tissue takes place

The Bulbus Cordis The bulbus cordis exists as a separate chamber in the heart of the fish and is provided with valves its function being to provide a means of regulation of the blood supply to the branchial system As the animal scale is ascended the necessity of a pulmonary circulation leads to its restriction to the right side In the embryonic human heart the bulbus forms the outflow portion and is situated at the right end of the common ventricle its extrapericardial portion being continued as the truncus arteriosus Within the bulbus are various

ridges and swellings which are described below and which are destined to form the septa dividing the bulbus In the human heart the bulbus is a transitory structure disappearing during the course of normal development and becoming mainly incorporated into the right ventricle to form its infundibulum or conus It disappears from the left ventricle its position being indicated by a small portion of the aortic conus The name of Keith (1909) is particularly associated with the bulbus and his work has greatly advanced knowledge of the morphology and fate of this particular structure in man Keith is of the opinion that the largest number of congenital heart anomalies arise as the result of arrest or interference in the normal process of involution of the bulbus Reflection on its role in the formation of the infundibulum and its contributions to the aortic and pulmonary valves and to other structures at once shows that it is a veritable keystone in congenital cardiac pathology Abnormalities of its incorporation lead not only to the different types of stenosis and hypoplasia of the pulmonary tract but also to transposition Its persistence in the left ventricle results in sub aortic stenosis

In the early stages of development the bulbus lies ventral to the primitive ventricle and the adjacent walls of the bulbus and ventricle become merged and then obliterated This allows free communication between the bulbus and right ventricle so that ultimately as the inter ventricular septum develops the bulbus is separated from the left ventricle and persists in the right ventricle to form the conus or infundibulum of the right ventricle The partitioning of the bulbus and its continuation the truncus arteriosus is a highly complex process The truncus arteriosus and bulbus exist initially as a continuous tube the line of division being the site where the aortic and pulmonary valves are formed Within this continuous tube two rows of swellings develop These are arranged in a spiral fashion and as they grow they unite to subdivide the channel into two approximately equal parts the aorta and pulmonary artery These ridges if traced downwards follow a spiral clockwise course Distally the ridges are laterally placed but as they course down towards the heart the right ridge curves on to the dorsal wall and then on to the left lateral wall The left ridge pursues a similar course in an opposite direction passing on to the ventral wall and then to the right wall In effect the septum formed by the fusion of these ridges has a twist in a clockwise direction of 270 nearly a complete rotation In this manner the posterior division of the tube comes to lie on the right hand side and the anterior division on the left Subsequent modification of this position places the pulmonary artery in front and the aorta behind The truncus septum or aortico-pulmonary septum fuses with the distal bulbar septum at the level of the semilunar valves The semilunar valves themselves are derived from four endocardial cushions in the distal portion of the bulbus The right

and left of these cushions fuse in such a manner as to form the distal bulbar septum and to leave in each half of the divided bulbus three cushions from which the semilunar valves are developed The distal bulbar septum joins the aorticopulmonary septum above and the proximal bulbar septum below The proximal bulbar septum is derived from the proximal bulbar swellings or ridges which are continuous with the right and left bulbar swellings so that when union is effected the bulbus is divided into aortic and pulmonary channels The fusion

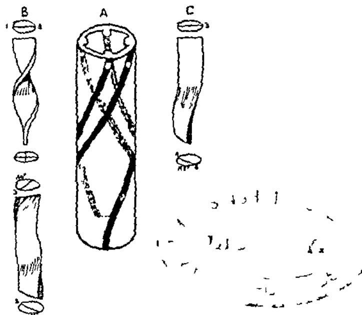


Fig 2 The bulbar septum A disposition of the bulbar ridges B normal absorption of bulbus C excessive back torsion about ridge A

of the proximal bulbar septum with the interventricular septum completes the division of the heart into a right and left side

The recent work of Pernkopf and Wirtinger (1935) suggests that there are two main stages in the development of the heart The first stage is concerned with the formation of the bulbo ventricular bend and the bulbar kink At the completion of this stage the bulbar ridges pursue a spiral course of 270 (fig 2)

The second stage is related to the absorption of the bulbus This process is necessarily a complex one and various twists or torsions take place in order to bring the different parts into their correct alignments Inspection of a normal heart shows that the pulmonary artery

arising anterior to the aorta undergoes a definite twist so that it passes behind the aorta on its way to the lungs Examination of the interior of the bulbus at the completion of the S shaped stage shows that the bulbar ridges pursue a helical course Such an arrangement of the bulbar ridges is present in the Dipnoean fishes and Robertson (1913) has indicated that kinking of the bulbus is sufficient to produce a spiral disposition of the ridges

Shrinking of the middle segment of the bulbus and notably that part of the bulbus embedded in the auricular furrow allows the formation of the aortic vestibule Linked with this process is an untwisting of the bulbar ridges and a transference of the twist to the ridges of the truncus this latter being shown by a torsion of 150 at the distal bulbar orifice Untwisting of the bulbar ridges is achieved by back torsion in *a counter clockwise direction at the proximal bulbar orifice Lev and* Saphir (1937) who have studied this torsion in relation to transposition state that this back torsion amounts to 45 and further leads to Ridge B *assuming a position further to the right For these authors the bulbo* auricular spur (or bulboventricular ridge of some authorities) constitutes a fixed point about which the bulbus rotates and torsion takes *place Ridge A which early joins with the ventricular septum may also* in some circumstances act as a similar fixed point

On this supposition is based Lev and Saphir s theory of transposition *Briefly if the bulbo auricular spur area is abnormal from any cause* it prevents the bulbus from embedding itself in the auricular furrow and a fixed point is not obtained In these circumstances untwisting of *the bulbar ridges is achieved by back torsion at the proximal bulbar* orifice in excess of that normally taking place The more detorsion at this site the less at the distal bulbar orifice so that in extreme degrees *of back torsion there is no torsion at the distal orifice Thus no twist is* given to the truncus ridges and the picture of transposition results Ridge A because of its early junction with the interventricular septum *may also act as a fixed point and rotation of the bulbus occurring* about it will again lead to the picture of transposition (fig 3)

Lev and Saphir s theory receives confirmation in alterations presented *by the muscle bundles of the infundibulum in specimens of* transposition examined by them These muscles are the septal and parietal muscles (left and right infundibular bands of Keith moderator band and crista supraventricularis of others) and are both readily identifiable in the human heart The crista supraventricularis a prominent crescentic muscle band constituting a boundary of the *pulmonary conus normally ends in the bulbar septum In cases of* dextroposition of the aorta with an interventricular septal defect it may be traced through the defect to the base of the aortic cusp of the mitral valve It is developed from Ridge B and the bulbo auricular spur and that portion passing through a defect may be interpreted as

a persistent bulbo auricular spur showing failure of atrophy in this region

Pulmonary stenosis and hypoplasia result from compression of the conus of the pulmonary artery between an abnormal bulbo-auricular spur and a septum developed by excessive back torsion about Ridge A A septal defect is chiefly the result of maldevelopment of Ridge 3 B Correct torsion brings Ridge 3 over Ridge B whilst detorsion tends to

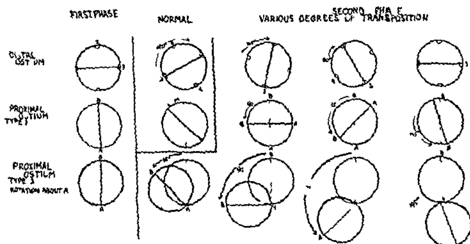


FIG 3 Diagrams of distal and proximal bulbar ostia in various types of transposition With increased back torsion at the proximal ostium either around its centre or round ridge A there is a lack of torsion at the distal ostium (Lev and Saphir)

place these structures apart Where there is improper formation of the aortic vestibule Ridge 3 B is under-developed and a septal defect occur

Lev and Saphir s work while differing in detail has confirmed the broad conclusions of Keith that transposition was the result of defective absorption of the bulbus It has also emphasized the integral part played by the bulbus in the early stages of cardiac development

The Atrial Canal This intervenes between the single auricle and ventricle of the primitive heart Four endocardial cushions appear within it and the anterior and posterior of these fuse to form the *auriculoventricular orifices* Up to this time the musculature of the auricle and ventricle is continuous but with the formation of the orifices connective tissue grows in from the atrial ring severing the muscular link between auricle and ventricle A few strands of muscle tissue escape and run down on the antero lateral wall from the sinus venosus These are destined to become the bundle of His

The atrial canal is a critical structure in the development of the heart because the interauricular and interventricular septa are both attached to the fused anterior and posterior endocardial cushions A failure in the development of the endocardial cushions will lead to abnormalities of septal formation and of the auriculoventricular orifices and valves

The Auricular Septum The septum primum appears at about the fourth week on the upper posterior part of the auricle and grows down

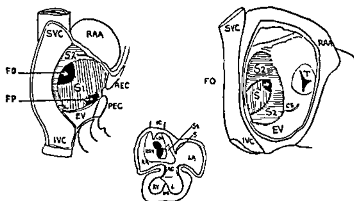


FIG 4 The formation of the interauricular septum (After Walmsley Quain s Anatomy) AC atrial canal AEC and PEC anterior and posterior endocardial cushions CS coronary sinus EV Eustachian valve FO fora men ovale FP foramen primum RSV and LSV right and left sinus valves S1 septum primum S2 septum secundum

wards towards the atrial canal Its lower margin is concave and when fusion of its extremities takes place with the endocardial cushions of the atrial canal a space the foramen primum is left above the atrial canal (fig 4) The upper posterior part of the septum primum disappears during the fifth week its place being taken by a septum secundum or foramen ovale In the meantime a second septum appears to the right side of the original attachment of the septum primum This is the septum secundum which grows down towards the atrial canal so that the two septa overlap and a slit like passage the foramen ovale intervenes between them The free edge of the septum primum in the left auricle persists as the valve of the foramen ovale The foramen ovale remains patent until birth when it may either completely close or persist as a slit like communication between the two auricles

Failure of development of the interauricular septum results in a persistent ostium primum persistent ostium secundum or a patent foramen ovale A persistent ostium primum is associated with anomalies of the auriculoventricular valves The septum may be absent

The Interventricular Septum Separation of the ventricles is necessary for the formation of an adequate pulmonary circulation The first indication of septation of the ventricles is the appearance in the fifth week of a ridge the septum inferius on the floor of the common ventricle Mall (1911) has shown that the growth of the septum from this point is not by upwards extension but by hollowing out of the primitive ventricular musculature to form the two cavities Dorsally the septum inferius fuses with the posterior endocardial cushion of the atrial canal

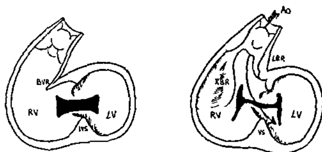


Fig 5 Separation of the aortic vestibule (Adapted from Gray s Anatomy 1932 25th Ed figs 188 189)

near its right end The septum inferius at this stage has a free concave upper margin

Between the auriculoventricular orifice and the proximal part of the bulbus cordis there is a bulboventricular ridge or spur (fig 5) corresponding with the external bulboventricular groove Fraser (1917) has shown that the disappearance of this spur allows the formation of the aortic vestibule During the formation of the septum inferius this ridge is absorbed with the result that the right end of the atrial canal comes to lie beneath the bulbar orifice The right and left bulbar ridges continuations of the right and left bulbar endocardial cushions together form the proximal bulbar septum The right bulbar ridge grows across the wall of the bulbus to the right extremity of the fused auriculoventricular cushions where it fuses with the free margin of the septum inferius obliterating part of the auriculoventricular orifice The left bulbar ridge grows along the free margin of the septum inferius until it meets and fuses with the right bulbar ridge As the proximal bulbar septum develops the ventral portion of the bulbus becomes incorporated in the right ventricle to form the infundibulum The dorsal portion of the bulbus almost entirely disappears although a vestige may remain in the aortic vestibule

The auricular septum fuses with the middle of the auriculoventricular cushions whilst the interventricular septum fuses with the right

extremity Thus between the right auricle and the left ventricle there is a small area derived from the fused cushions This becomes the mem branous portion of the ventricular septum

Failure of development of the ventricular septum is closely linked with the involution of the bulbus and abnormalities of torsion as discussed above Most defects of the interventricular septum are conse quently associated with other cardiac anomalies The isolated defect of the ventricular septum is generally anterior to the membranous septum and due to arrest of development of unknown origin It is rarely due to a persistent interventricular foramen It may be due to a slight dextro position of the aorta a minimal form of transposition Defects towards the apex of the ventricular septum have a morphological interest as such defects are found in the heart of the python

Morphology

Darwin (1859) stated that the metamorphosis of the individual recapitulated the evolution of its kind A remarkable feature of con genital cardiac pathology is the resemblance presented by the deformed human heart to the heart of the various classes of the lower vertebrata A knowledge of these hearts is of help in the identification of the grosser human cardiac defects

The heart in the lower vertebrates is a relatively simple muscular organ designed to drive venous blood forward to the gills Originally in the lowest animal orders a simple straight cylindrical tube it becomes differentiated in the vertebrata into four chambers each more or less separated by a constriction If one of the lower fishes (fig 6) is taken as a type the heart is roughly bilaterally symmetrical The most posterior of the chambers is the sinus venosus into which enters the ducts of Cuvier and hepatic veins and also blood from the heart itself by a cardiac vein The impulse originates in the sinus venosus and the blood is propelled forward by rhythmic contraction passing successively over the chambers This mechanism is of significance for in the human heart the impulse arises in the sino-auricular node A venous valve of two cusps intervenes between the sinus and the common auricle The latter is thin walled and very distensible and owing to a slight bending of the cardiac tube is situated somewhat dorsal to the common ventricle The common auricle communicates with the large thick walled ventricle which tapers towards its anterior end to become continuous with the bulbus arteriosus or bulbus cordis The bulbus cordis which in the fish has the status of a separate chamber gives off the branchial arches at its extreme anterior end The development of the bulbus varies in the different orders of fishes In the Elasmobranchs (sharks and rays) it is well developed and contains several rows of valves in the Teleosts it is considerably reduced and may bear only one row of valves in the Dipnoi (mud and lung fishes) where there is a double respiration by

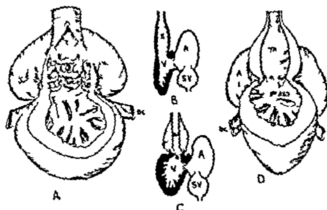


Fig. 6 The heart of the fish A, B elasmobranch C, D teleost (B and C ex Quain's Anatomy 1929 Vol. VI part 3 page 8 fig. 3)

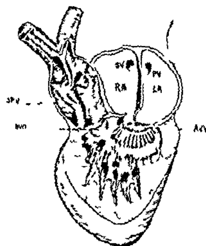


Fig. 7 The heart of the frog AVV auriculoventricular valve BVO bulboventricular orifice SPV spiral valve

lungs as well as gills the bulbus is spirally twisted upon itself an partially divided into two channels by fused valves thus representing stage in cardiac development between the fishes and the amphibia In addition in the Dipnoi the auricle is divided by an interauricula *septum and the sinus venosus opens into the right atrium*

In the amphibia certain changes take place in view of the developmen of a pulmonary circulation and the disappearance of the gills (fig 7) The heart acquires the shape characteristic of the higher vertebrates and the S shaped curvature of the heart tube is such that the auricle is carried dorsal to the common ventricle so that the latter opens back wards into it The sinus venosus is also carried dorsally and opens *downwards into the auricle The auricle is partially subdivided into a* large right and a small left auricle by a crescentic *interauricular septum* the free margin of which is above the auriculoventricular orifice The left auricle receives a pulmonary vein and the sinus venosus opens into

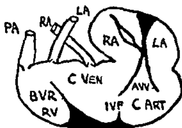


Fig 8 Diagram of reptilian heart (After Quain) AVV auriculoventricular valves BVR bulboventricu lar ridge above which rises the left aorta LA C VEN and C ART *divisions of the dorsal ventricle* RV *right* (ventral) ventricle

the right auricle The sinus venosus still remains a separate chamber receiving the ducts of Cuvier the inferior vena cava and a coronary vein The bulbus is spirally twisted and contains a spiral valve but is however incompletely partitioned Small coronary arteries are derived *from the first arch*

In the reptilia (fig 8) the heart is more specialized and is divided into two sides by the auricular and ventricular septa The interauricular septum meets the auriculoventricular orifice and divides it In the higher types the sinus venosus is more or less incorporated into the right auricle *but in many species it remains an actual chamber guarded by a sinus valve The pulmonary veins open into the left auricle* The ventricle is divided into two chambers a small anterior and a large posterior by an interventricular septum with a free concave upper margin in the lizard snake and tortoise In the crocodile the interventricular septum

is complete A right and left aorta are in relation to the posterior or left ventricle and a pulmonary artery springs from the anterior or right ventricle All three great vessels are furnished with a bicuspid valve A feature of the reptilia is that the bulbus no longer exists as a separate chamber and has become submerged in the ventricle It has undergone a spiral twist and by fusion of its valves been partitioned into the two aortae and the pulmonary artery

In the bird the septa are completely formed The bulbus has disappeared and both aorta and pulmonary artery bear three cusps The sinus venosus is incorporated in the right auricle The tricuspid valve consists of three thick muscular folds and the mitral valve has three cusps The aorta crosses the right bronchus

It is thus possible to trace the general evolutionary lines of the heart (fig 9) It begins as a simple muscular enlargement designed to pump blood to the gills Later it is subdivided into four chambers protected by valves in order to ensure the forward flow of the blood Beginning

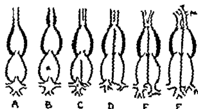


FIG 9 Diagram showing the heart in different vertebrates A elasmobranch B teleosts C amphibia D lower reptiles E alligator F birds and mammals a auricle b bulbus cordis s sinus venosus v ventricle (After Kingsley)

with the fish it gradually acquires an S shaped bend and valves appear in the bulbus cordis As the scale is ascended and the transition occurs from the gill breathing to the lung breathing types the heart is divided into two sides for the purposes of a pulmonary and a systemic circulation Definite subdivision of the auricles takes place in the amphibia and important subdivision of the ventricle in the reptilia becoming complete in the crocodilia The bulbus cordis ceases to exist as a definite separate and functional chamber after the amphibia but is present in the embryonic state of all the vertebrates It first becomes spirally twisted and partially subdivided in the Dipnoean fishes An incomplete spiral septum divides the bulbus in the amphibia This becomes complete in the reptilia where the bulbus is partitioned into three compartments comprising two aortae and a pulmonary artery Above the reptilia there is a single aorta and pulmonary artery

Table summarising the principal points in the morphology of the heart in the vertebrata

	SINUS VENOSUS	AURICLE	VENTRICLE	BULBUS CORDIS	AORTA
FISHES	Opens into the atrial cavity with an intervening valve	Undivided	Undivided	Separate chamber	
Dipnoi	Opens into R atrium	Right and left	Undivided	Spiral twist and incomplete fusion of valves	
AMPHIBIA	In some species sunk in the R auricle	Well developed septum May be fenestrated	Undivided	Separate chamber incompletely di vided by spiral septum	Coronary arteries from 1st arch
REPTILIA	Not a distinct chamber in most species	Divided Single A V cusps	Incomplete except in the Crocodilia	Not a separate chamber Divid ed into R and L aorta and pul art Each with two cusps	Two aortae
BIRDS	Scarcely recogniz able	Divided R AV valve muscular L three cusps	Divided Papillary muscles		Right aortic arch
MAMMALS	Eustachian and Thebesian valves	L AV valve bi cuspid		Mainly incorpor ated in the R ventricle	Left aortic arch

CHAPTER III

FREQUENCY, ETIOLOGY CLASSIFICATION

Frequency

It is difficult to arrive at any exact figures relative to the incidence of congenital heart disease The recognition of congenital heart disease is to some extent proportional to the knowledge and interest of the observer and it is only within the last few years owing to the pioneer work of Abbott (1927) and the more recent advent of surgical treatment that any deep interest in the subject has been generally displayed Previously it was considered unwise to venture beyond the mere diagnosis of congenital heart disease probably because little could be done in the way of treatment, and many cases especially in the acyanotic groups escaped etiological recognition Even to-day despite the implications of surgery there is some hesitancy in diagnosis although a large volume of clinical and other evidence permits reasonably accurate recognition of a number of anomalies There remains however a singular dearth of statistical evidence of incidence such as is available falling into two main orders post mortem and clinical A mass of autopsy evidence mostly from American sources is gradually accumulating owing to impetus given to the matter by Abbott (1936) but there is little of such work from British sources The more recent information is summarized in the table below

TABLE I (POST MORTEM INCIDENCE)

	Total Autopsies	*Cases of Congenital Heart Disease*	*Percentage of Cases*
Rannels and Propst	4 255	36	0.85
Szypulski	7,500	111	0.17
Roberts	27 077	107	0.009
Dolgopol	1 09[illegible]	64	5.9
McGinn and White	7 500	67	0.09
Philpot	7 240	80	1.1
Leech	13 115	170	1.29
Nicholson	1 851	37	2.00
Terplan and Sanes			
(children under three) Buffalo	—	—	5.4
Prague	—	—	1.3
De la Chapelle	8 683	31	0.36
Jacobius and Moore	1 600	131	8.1
Ingham (Mayo Clinic)	8 314	87	1.05

From these figures it appears that the average incidence in all post mortems is in the neighbourhood of 2 per cent As some of these figures are derived from children s hospitals and include neonatal cases the figure should be generally somewhat lower

Clinical statistics are equally scarce and are again mostly derived from sources other than British This is surprising because there exists in the British Isles a school medical service whose responsibility it is to examine all school entrants at the age of five Heart cases are how ever only segregated into organic and functional and only where special heart clinics are conducted by the various authorities are any data available Even so this information is difficult of access and must be sought in the yearly reports published by the several authorities White (1931) summarised the then available literature of the incidence of heart disease according to etiology The figures below are derived from this source

TABLE II

	Cases of Congenital Heart Disease	*Percentage of cases*
White and Jones (1928)	1 543	2 5
Wyckoff and Lingg (1926)	1 001	05
Wood Jones and Kimbrough (1926)	300	1 0
Stone and Vanzant (1927)	915	1 0
Myers (1927)	510	2 0
Willius (1926)	330	1 0
Viko (1930)	867	1 0
Coffen (1929)	1 344	0 1
Coombs (1926)	1 000	2 0
Parkinson and Clark Kennedy (1926)	100	1 0

From these figures it appears that between 1 and 2 per cent of cases of organic heart disease are of congenital etiology It seems probable that with the continual advances in the recognition of these cases by clinical methods this figure may slightly increase

Etiology

From an etiological point of view congenital abnormalities of the heart fall into two main groups The first group comprises those cases due to a primary arrest or defective development occurring before the eighth week of intra uterine life The second group includes those rare cases which are the result of infection transmitted by the mother to the foetus after the second month when the septa have closed

Hope (1839) believed that the commonest cause of heart anomalies was an arrested development Bouillaud (1835) recognised the dual origin by arrest of development or infection Defective or arrested development is responsible for by far the greater number of congenital heart cases and most experts would now doubt the importance

originally attributed to foetal endocarditis What other factors may cause errors in development remain largely a matter of conjecture

A study of any large series of congenital heart defects will reveal the not inconsiderable frequency with which there are defects elsewhere in the body and dispel any doubt that their coincidence is accidental Figures of such an association vary within wide limits from the extreme of 54 per cent (Jones 1926) to 18 per cent (Abbott 1927) and 11 per cent (Vierordt 1898) Brumlik (1937) noted 6 per cent in clinical cases and 23 per cent in autopsied cases Such associated visceral or somatic abnormalities may be transposition of the viscera cleft palate hare lip poly and syndactyly congenital hernia spina bifida and others In our experience one or more accessory nipples appear to be the commonest of all associated congenital abnormalities

Chest deformities are quite common irrespective of any history of infantile asthma pneumonia bronchitis or rickets Attention has been called to the frequency of Harrison s grooves in severe grades of con genital heart disease by Naish and Wallis (1948) Little is known of the exact causation of these sulci Sheldon (1938) suggested that pulmonary atelectasis occurring as a complication of heart disease probably caused indrawing of the intercostal spaces

Mongolism and arachnodactyly are of especial interest owing to the frequency with which congenital heart disease is found in these con ditions The causes of mongolism are at present unknown and it is said to occur as an exhaustion product when maternal age is advanced or at the end of a large family That this is by no means always the case has been shown by Still (1927) (fig 10) and it is within the experience of most physicians to observe the first child of a family thus affected It is only rarely that more than one mongol occurs in a family Maternal age is still held by many to be a most important factor and both hormones (Brousseau 1928) and vitamins have also been incriminated as etiological factors The work of Meyer and Cook (1937) shows that in some cases there is a fibroglial sclerosis involving chiefly the white matter which may possibly be due to anoxaemia It at least appears to be certain that the factors producing mongolism operate in the early weeks of intra uterine life at a time when the development of the heart may be influenced The study of mongolism may well throw light upon the genesis of cardiac defects The general characteristics of mongolism are so well known that the condition may be recognized at a glance Suffice it to say that the extremities are usually cold and cyanosed owing to poor circulation even in the absence of a congenital heart lesion The frequent association of congenital heart disease with mongolism has long been recognized Figures of the incidence of con genital heart disease in mongols have been given by Thompson (1920) who found heart signs in one of every five examined Other statistics have been contributed by Thursfield (1926) who found heart disease in

16 per cent Spuhler (1929) 25 per cent Edelhaus (1932) 21 per cent Abbott (1927) considered that the defects of the atrial septum and in particular a defect of the septum primum were the commonest abnormalities present in mongols Silvy (1934) found signs of the maladie de Roger in 61 per cent of fifty five mongols with heart disease

In arachnodactyly the incidence of congenital heart disease is perhaps higher than in the mongol Arachnodactyly was first described by Marfan (1896) and is an hereditary disease The most prominent

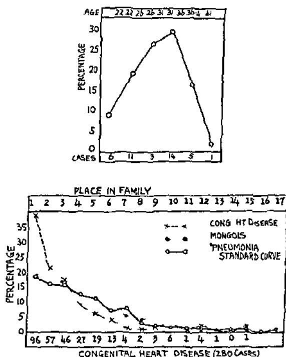


FIG 10 Showing age of mother at birth of infant with congenital heart disease and place in the family of congenital heart disease and mongols (From G F Still)

signs are abnormally long slender fingers and toes The long bones are also involved and the subjects are tall and underweight for their age and have a general appearance of emaciation owing to poor musculature and lack of subcutaneous fat The joints show excessive mobility owing to relaxation of the ligaments Associated with the skeletal abnormalities there may be deformities of the feet kyphosis and scoliosis or deformities of the sternum Other striking features that may be present are dislocation of the lens high arching of the

palate and prominent ears A congenital abnormality of the heart or aorta is present in a large number of cases Rados (1942) analysed the literature up to 1940 and collected 204 cases cardiovascular abnormalities being present in sixty three Auricular septal defects occurred in three of four cases of Piper and Irvine Jones (1942) and interventricular septal defects patent ductus arteriosus aortic regurgitation and cardiac enlargement have all been mentioned An interesting associated anomaly is medionecrosis of the aorta occasionally accompanied by dissecting aneurysm in some instances terminating in sudden death from rupture The presence of medionecrosis of the aorta or of the commonly associated vascular hypoplasia renders surgery undesirable Such are the cases of Baer Taussig and Oppenheimer (1943) Etter and Glover (1942) and Uyeyama Kondo and Kamins (1947) The rheumatic infection has been quite often associated with arachnodactyly The etiology of arachnodactyly remains obscure Some have considered it to be a congenital mesodermal dystrophy owing to the multiplicity of structures that may be involved Whatever its exact etiology may be it seems clear that the factors in its production operate early in foetal life at a time when the heart may be influenced Ellis and van Crefeld (1940) have described a syndrome characterized by ectodermal dysplasia polydactyly chondrodysplasia and congenital heart disease

The occurrence of congenital heart disease in monsters has only as yet been partially studied In the embryological sense the term monster designates a foetus or full term child in which malformations are present whether of the heart spine or other organ Mall (1917) stated that monsters were not due to any hereditary influence but were due to factors in the environment of the foetus The tendency towards the predominant involvement of one system was thought by him to be evidence that different tissues were affected at critical stages in their development He found that most monsters developed before the eighth week Scammon (1925) stated that monsters could be produced by certain chemical or physical agencies such as extremes of temperature X rays and chemical changes in the surrounding fluid of the developing embryo These experimental and environmental matters have been discussed by Reid (1931) Keith found a malformation of the heart in fourteen of twenty three malformed foetuses and a personal unpublished series shows a high incidence of cardiac anomalies in spina bifida anencephaly and other severe malformations Mall found monsters to be twelve times as frequent amongst abortions as in full term births The causes of abortion are well known and include pathological conditions of the chorion and amnion uterine disease imperfect implantation of the ovum endocrine disturbance and avitaminosis In regard to this latter it has recently been shown (Warkany 1944) that female rats fed and maintained on a diet deficient in vitamin A produced

young with multiple congenital defects These defects principally involved the eyes but there were also defects of the lungs and pleura abnormality of the heart muscle and in a number of instances a right sided diaphragmatic hernia As liquid paraffin hinders the absorption of Vitamin A and is frequently used as a laxative in pregnancy its continued use may not prove to be desirable All the above can give rise to important environmental changes in relation to the developing foetus

Thomson (1920) is of the opinion that disturbance of the health of the mother in the early weeks of pregnancy might have an important influence upon the production of malformation

Gregg (1941) in Australia reported that certain congenital abnormalities affecting the ear eye and heart were related to a maternal attack of rubella in the early months of pregnancy Of seventy eight cases with congenital cataract forty four had congenital heart disease This received a somewhat mixed and sceptical reception for after all why had not this association been noted before particularly as a rash in pregnancy could hardly escape observation? Likewise why should such a trivial disease as rubella produce such devastating effects upon the embryo when other and more severe infections left the foetus unscathed? Confirmatory reports soon began to appear Swan (1943) and others found in a series of maternal rubella in pregnancy that thirty one children of forty nine mothers had congenital abnormalities in seventeen involving the heart Reese (1944) noted a similar association in three further children with congenital heart disease These reports could be substantially multiplied In a personal (unpublished) case maternal rubella occurred in the fifth week of pregnancy The infant born at term had an interventricular septal defect and congenital cataracts

Interrogation of the mothers of 400 congenital heart cases in a personal series has failed to reveal a single instance of rubella or of any other infectious disease in the early months of pregnancy One is entitled to ask if a mother would remember what is usually considered to be a trifling illness often treated without the aid of a doctor Further could the rubella of Australia be a new form of the disease or one enhanced in virulence by the importation of service personnel from overseas? Certain ectodermal structures such as the lens nucleus are well known to be vulnerable to virus infection but it is difficult to understand how mesodermal structures such as the interventricular septum can be involved unless by interference with its vascular supply or infiltration of its developing tissues Brown (1947) considers that the adrenal is the principal structure attacked by the virus leading to adrenal failure which in its turn leads to defects in the eyes heart ear teeth and brain This view is open to criticism because the first steps of development of the cortex and medulla are only apparent in the fifth

and sixth week when the cardiac septa are all but formed and there is no evidence that they are functioning organs apart from this The subjects of congenital heart disease supposedly initiated by rubella have not been demonstrated to be cases of simultaneous hypoadrenalism The association of arachnodactyly congenital cataract and defect of the auricular septum might now bear investigation in relation to maternal infection

The elucidation of the problem of rubella is important Unfortunately the records of maternity clinics and of most heart clinics will be of little use as hitherto rubella has not been looked upon as a significant disease The best approach will be the examination of the children of mothers definitely known to have had infectious diseases in the early months of pregnancy The fact does remain that rubella in pregnancy must be looked upon as a formidable disease Mothers exposed to infection with rubella or mumps might well receive an injection of the gamma globulin fraction of convalescent serum as soon after exposure as possible Rubella in the first three months of pregnancy should be considered to be an indication for therapeutic abortion

HEREDITARY INFLUENCES Inherent changes in the germ plasm have been invoked as a possible cause of defect Comparatively few cases of congenital heart disease attain maturity and fewer still reproduce so that any assessment of the importance or otherwise of this factor remains almost impossible in the present state of knowledge The reported cases mainly suggest that heart anomalies are more often found in members of the same generation than in the ancestors Abbott (1927) found a heart anomaly in brother or sister in eleven cases Patent ductus arteriosus has been found in twins (Smith 1929 Brown unpublished) (fig 42) in sisters (Jewsbury 1912 Ellis 1933 Snelling 1937) Congenital idiopathic hypertrophy has occurred in sisters (Sprague Bland and White 1931) the maladie de Roger in mother and child (Debre 1923) septal defects in a father and four children (Seitz and Baumann 1935) congenital heart block in father and child (Walgren and Winblad 1937) and in two sisters (Aitken 1932) A personal (unpublished) series shows congenital heart disease in brother or sister in six instances (fig 43) On the other hand in dextrocardia a defect of no clinical significance its occurrence in four generations was noted by Lancisi (1701) and in three generations by Muir (unpublished) In all the literature contains sixty three reports of congenital heart disease in two or three members of a family (Courter *et al* 1948) The genetics of transposition of the viscera and dextrocardia have been studied by Cockayne (1938) Rosler (1928) observed consanguinity in 10 per cent of cases of cardiac abnormality

Murphy (1940) considers that the factors responsible for the development of a congenital defect are present in either the sperm or the ovum before fertilization and if this view is correct there must always be a

certain number of congenitally deformed individuals occurring at a more or less constant rate The age of a parent at the birth of its off spring is believed to be important after the age of 30 and after a maternal age of 40 the birth rate of malformed individuals is three times the rate of those born when the mother is less than 30 It is also thought that the parents of a malformed child are likely to have a further abnormal child A gross malformation is stated to occur once in every 200 live births amongst the population at large whereas in the offspring of the parents of a malformed child another malformed child is likely in every eight births Therefore from a preventative point of view only the limitation of reproduction after the age of 30 and no further reproduction by the parents of malformed children is likely to have any effect upon the frequency of abnormality How far this view point is applicable to congenital heart disease must remain a debatable problem in the light of our present knowledge

The inference to be drawn is that in so far as congenital defects are concerned the environmental conditions of the foetus play a prominent part The health of the mother is a factor of considerable importance but one that is not always amenable to supervision because the case of pregnancy rarely comes under observation until after the third month at a time when efforts directed towards the prevention of a cardiac defect are useless Rubella in the early months of pregnancy is of great significance It seems worth while to consider the advisability of attempting to save a threatened abortion in view of the damage that may be sustained by the foetus and the major incidence of anomalies in abortions

FOETAL ENDOCARDITIS Infection transmitted by the mother to the foetus after the heart has reached its definitive form and the septa have closed has long been accepted as a cause of congenital cardiac path ology As early as 1814 Kreysig thought that cases of pulmonary stenosis with a patent foramen ovale were the result of a foetal endo carditis Similar views were entertained by Laennec (1837) who in describing a case where the valvular orifices were generally narrowed stated that such cases were occasioned by une inflammation des valvules qui aurait eu lieu chez le foetus Rokitansky (1842) thought that the foetal endocarditis had a predilection for the right side of the heart whilst *endocarditis in post natal life more frequently* involved the left side Peacock (1866) states the pulmonary valves are often found united in cases in which the septum of the ventricles is imperfect or where the foramen ovale is unclosed and the ductus arteriosus still pervious conditions which clearly point to the existence of obstructions at the pulmonary orifice during foetal life It may be inferred that the disease processes on which they depend are in their *nature pre* cisely similar to those which give rise to thickening and adhesions in after life Peacock also observed the major incidence of foetal inflam

matory processes in the right side of the heart and attributed this to the more intimate connection of the right ventricle in the foetus with the circulation in the descending aorta and umbilical arteries Because he considered variation in intra arterial pressure in the aorta to be causative of the aortic valvular disease by analogy he thought that interference in the circulation in the placenta and the cord by leading to fluctuations in pressure in its turn might explain the increased liability to disease of the right side of the heart in the foetus The theme of foetal endocarditis was subsequently amplified and for a long time it was a well recognized etiological factor in certain lesions Increase in knowledge and experience has largely displaced foetal endocarditis from its former role of importance As a matter of clinical observation it is uncommon to observe cases where a congenital heart lesion can be correlated with infection in the mother The coincidence of the rheu matic infection in an active stage with pregnancy is comparatively rarely noted Cases do however occur where cause and effect are evident The result of infection is a foetal endomyocarditis This terminates in thickening and distortion of the valve cusps and gives rise to lesions of which the types are the valvular atresias and pul monary stenosis with a closed interventricular septum Examination of such a heart will often show its true pathology at a glance although in some cases microscopical examination may be necessary The latter is often necessary in cases of apparent bicuspid aortic valve to separate those which have a true developmental origin and those which have arisen as the result of inflammatory process The macroscopic appear ance of the valve may be similar to that found in the chronic valvulitis of the adult If the inflammation has resulted in severe stenosis of the valve amounting to atresia there will be other changes of importance of a secondary nature In aortic atresia there is hypoplasia of the aorta and left ventricle In an aortic stenosis the left ventricle is hypertrophied and dilated Scarring may be evident in the walls of the ventricle giving the endocardium a pearly white appearance Calcification may be present and the presence of a myocarditis is constantly confirmed by microscopical examination The subject of foetal endocarditis has been well studied by Farber and Hubbard (1933) The influence of other infections is problematical As regards syphilis which is particularly incriminated in continental schools the usual lesion is myocardial and consists of a fibrous myocarditis with numerous colonies of spiro chaetes Warthin (1911) has shown and Farber and Hubbard endorse the view that valvular lesions are conspicuously absent in congenital syphilis Whatever infection may be the basis of a gross anatomical defect a certain amount of the changes in the myocardium are of a secondary nature and may be ascribed to a deficient blood supply to the heart muscle producing fibrous tissue replacement and other myocardial lesions These changes may be observed in aortic atresia

and similarly in those cases where a coronary artery arises from the pulmonary artery

SEX INCIDENCE In Abbott s (1927) series the males are in excess of the females the ratio being about 4 3 In a series of 300 cases (unpublished) observed by Brown and Muir the females are slightly in excess of the males There are however certain peculiarities in the distribution of the cases between the sexes at once discernible if any long series is reviewed Simple patency of the ductus arteriosus is nearly twice as frequent in the female as in the male while the Lutembacher syndrome (mitral stenosis and interauricular septal defect) appears to be almost exclusively found in the female The uncomplicated auricular septal defect is commoner in the female A bicuspid aortic valve coarctation of the aorta aortic stenosis and atresia defects of the aortic septum and congenital aneurysm of the right aortic sinus of Valsalva are all much commoner in the male In the cyanotic groups the incidence in the sexes is about equal but in more complicated anomalies such as the cor biloculare and cor triloculare the incidence in the male exceeds that in the female

Congenital heart disease may be encountered at any age but naturally is more common in infants and young children As many cases die in infancy or early childhood the number diminishes as age advances There still remain many who survive until adult life particularly in the acyanotic groups Some cases may attain a comparatively old age The race to which the patient belongs does not influence the incidence as far as can be ascertained

Classification

Several classifications of congenital heart disease have been proposed The importance of cyanosis has long been recognized and its existence or non existence or its occasional occurrence has been used by Abbott and Dawson (1924) as a basis for their well known groups Their classification is as follows

(A) OF CLINICAL SIGNIFICANCE

I NON CYANOTIC GROUP

(1) Cases in which no abnormal communication exists but in which the defect may produce a mechanical interference with the circulation and become the seat of strain

- Pericardial defect
- Ectopia cordis
- Primary idiopathic hypertrophy
- Congenital heart block
- Anomalous septa
- Pulmonary and tricuspid insufficiency

Aortic and mitral stenosis
Anomalies of semilunar and auriculoventricular cusps
Coarctation and hypoplasia of the aorta
Anomalies of the aortic arch
Congenital arteriovenous aneurysm
Anomalies of veins and of the pulmonary artery

(2) Cases of arterial venous shunt with possible transient or terminal reversal of flow (cyanose tardive)

(*a*) Defects of the auricular septum
Persistent ostium primum
Persistent ostium secundum
Patent foramen ovale
Multiple defects
Complete defect Cor biventriculare and triloculare

(*b*) Localized defects at the base of the interventricular septum
Maladie de Roger

(*c*) Patent ductus arteriosus

(*d*) Localized defects of the aortic septum

(*e*) Congenital aneurysm of the right aortic sinus of Valsalva

II CYANOTIC GROUP

(*a*) *Moderate Cyanosis*

Dextroposition of the aorta with defect at the base of the interventricular septum
Complete absence of interventricular septum
Pulmonary stenosis with closed septum
Tricuspid atresia with atrial septal defect and transposition of vessels

(*b*) *Marked Cyanosis*

Tetralogy of Fallot
Pulmonary atresia with ventricular septal defect
Transposition of trunks with ventricular septal defects

(*c*) *Extreme Cyanosis*

Cor biloculare
Persistent truncus arteriosus
Transposition with closed ventricular septum
Pulmonary atresia with closed septum
Aortic and mitral atresia

(B) Of No Clinical Significance

Acardia
Dextrocardia

Dry (1937) has proposed a clinical classification based on broad embryological grounds It is of interest and represents a new approach to the subject

His classification is as follows

I Anomalies associated with septal formation
- Cor biloculare
- Cor triloculare biatriatum
- Auricular septal defects
- Ventricular septal defects
- Persistent truncus arteriosus

II Anomalies associated with torsion of the cardiac tube and

III Anomalies associated with the development of the bulbus cordis
- Subaortic stenosis
- Pulmonary stenosis
- Transposition of the great vessels
- Tetralogy of Fallot
- Eisenmenger s Complex
- Anomalies of the aortic and pulmonary valve cusps

IV Anomalies associated with the development of the aortic arches
- Persistent right aortic arch
- Double aortic arch
- Coarctation of the aorta
- Anomalous origin of vessels or the arch
- Patent ductus arteriosus

V Dextrocardia

VI Anomalies of the coronary vessels

Sussman s (1946) classification has a great deal to recommend it as *it was devised primarily for radiologists* and concerns those cases which are compatible with life and therefore of interest to radiologists and clinicians It does not comprise the perhaps larger group of cases of interest to pathologists Sussman arranged the abnormalities as follows

1 *Obstruction of the Outflow Tracts*
- Isolated pulmonary stenosis
- Aortic or subaortic stenosis
- Coarctation of the aorta

2 *Intracardiac Shunts*

Isolated interventricular septal defects (Roger)
Auricular septal defect
Tetralogy of Fallot
Eisenmenger s Complex

3 *Extracardiac Shunts*

Patent ductus arteriosus
Arteriovenous aneurysm

4 *Miscellaneous Anomalies*

Positional changes in the heart and great vessels (e g dextro cardia and transposition of the vessels)
Idiopathic dilatation of the pulmonary artery
Congenital aortic aneurysm
Aberrant pulmonary veins with atrial septal defect
Anomalous origin of coronary artery with left ventricular dilatation and hypertrophy
Congenital hypertrophy from which von Gierke s disease must be distinguished

The classification of Abbott and Dawson is mainly adhered to throughout this work

CHAPTER IV

THE ACYANOTIC GROUP

Pericardial Defects and Congenital Idiopathic Hypertrophy

This group comprises those cases where there is no abnormal communication between the two sides of the heart The anatomical lesion present may however be the cause of strain or of lowered resistance

The principal members of this group are those anomalies termed by Laubry and Pezzi (1921) left sided lesions These are aortic and sub aortic stenosis mitral stenosis coarctation of the aorta hypoplasia of the aorta and anomalies of the aortic cusps A peculiar feature of the group as a whole is the frequency with which the lesions may be com bined in the same individual Examples of such an association are aortic and subaortic stenosis and the better known combination of coarctation and a bicuspid aortic valve

Other members of the group are pericardial defects and anomalous bands and chordae A right sided aortic arch finds its natural place here and congenital arteriovenous aneurysm is also included in this general group

Defects and Abnormalities of the Pericardium

Historical Note Absence of the pericardium was first reported by Realdus Columbus in 1559 but in all probability this was an adherent pericardium Baillie (1793) gives a detailed description of a case encountered in the dissecting room Many of the early cases are un doubtedly examples of adherent pericardium Southworth and Stevenson (1938) could find but fifty two undoubted cases seven of these being monsters Since then cases have been reported by Dahl (1937) Osgood and Spector (1941) Ronka and Tessmer (1944) and Sunderland and Wright Smith (1944)

Embryology and Pathogenesis

(1) Development of the Pericardium

The earliest traces of the pericardium are visible in the 1 5 2 mm embryo At this time the mesoderm is hollowed out to form the primi tive pleural pericardial and peritoneal cavities The pericardial cavity itself is formed by the fusion of spaces in the mesoderm surrounding the heart A bank of mesoderm the septum transversum intervenes between the developing pericardium and peritoneum and contains the

developing liver The two cavities remain in communication by the dorsally situated paired pericardial peritoneal canals The developing lung buds ultimately herniate into these canals to form the pleural sacs

At about the fifth week the pleural sacs become separated from the pericardial and peritoneal cavities At this time the ductus Cuvieri comes into contact with the pericardio peritoneal canal and compresses it as it passes from the body wall to the heart Pressure on the canal raises a pulmonary ridge which reduces its lumen The cranial end of the ridge the pleuropericardial membrane is closely related to the opening between the pleural and pericardial cavities As these cavities enlarge the connection between them is reduced to a minute foramen Closure is effected not only by the active growth of the cavities but also by the combined effects of pressure exerted by the growing ductus Cuvier and the continued enlargement of the pleuro pericardial membrane

Sunderland and Wright Smith have examined the factors which might be held to be responsible for a pericardial defect These acting singly or in combination have been summarized by them as follows

(1) The normal stretching of the pericardium coincident with enlargement of the heart

(2) Premature and/or abnormal enlargement of the heart which precedes the closure of the pleuro pericardial foramen Projection of the heart through the opening at this stage precludes any possibility of closure at a later date

(3) An abnormally large foramen not fully closed when heart and pericardium begin to enlarge

(4) Delayed or non closure of a normal pleuro pericardial foramen which is subsequently enlarged as the pericardium stretches to accom modate the developing heart This arrested development may be due to defective formation of the pleuro pericardial membrane owing to premature obliteration of the duct of Cuvier or failure of the differential growth factor which reduces the foramen between the pleural and pericardial cavities to a minute foramen

(5) Failure of a pleuro pericardial membrane whose development has been delayed to grow sufficiently rapidly to close a defect which is being slowly extended by the expanding pericardium containing the enlarging heart

It would appear then that abnormal development of the pleuro pericardial membrane is a key process in the genesis of abnormality of the pericardium and that a contributing factor of importance is the tendency of a growing heart to maintain stretch and even herniate into an opening in its enveloping sh ath The predominance of abnor mality on the left side is due to changes in the duct of Cuvier and enlargement of the heart to the left

PATHOLOGY Abnormalities of the pericardium can be divided into distinct groups

(1) Complete absence of the parietal pericardium the heart and left lung occupying a common pleuropericardial cavity 76 per cent of cases (Southworth and Stevenson)

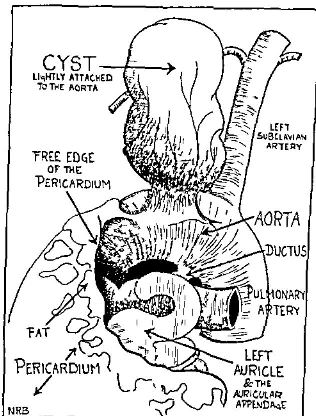


FIG 11 Deficient pericardium and pleuropericardial cyst Male aged 49 (Mr N R Barrett s case)

(2) A defect or foramen between the pericardium and left pleural cavity the heart occupying the pericardial sac 24 per cent of cases

(3) Complete absence of the parietal pericardium the heart lying in its normal mediastinal position *Probably never occurs (Southworth and Stevenson Sunderland and Wright Smith)* The reported cases are in all probability examples of adherent pericardium It has been described in dogs by Schlotthauer and Stalker (1936)

Pericardial defects may be associated with defects of the heart or

of other viscera or occur in monsters The defect is three times commoner in the male than in the female

The heart may be normal in size but in about half the reported cases it was enlarged in some cases owing to hypertension valvular or other disease In other cases enlargement may be present without

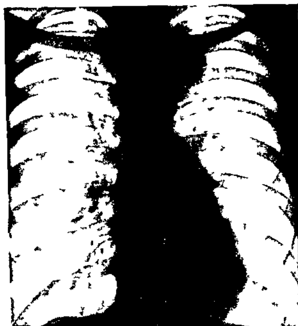


FIG 12 Deficient pericardium and pleuropericardial cyst Male aged 49 Cyst successfully removed and patient in good health (Mr N R Barrett s case)

adequate explanation The left of the parietal pericardium is almost invariably involved the extent of abnormality varying from a small foramen to a virtual complete absence of the left leaf In the case of Sunderland and Wright Smith the remaining pericardium presented a free margin running transversely across the heart leaving the anterior surface mainly uncovered A similar example is the unpublished case of Barrett (figs 11 and 12)

CLINICAL PICTURE Absence of the pericardium or a part of it rarely gives rise to clinical signs and symptoms and discovery of a case has generally been fortuitous or in the post mortem room Ladd (1936) encountered the defect during the repair of a diaphragmatic hernia in

a child of 2 Dahl (1937) produced a pneumopericardium during the establishment of a pneumothorax and thought that a pericardial defect was the only explanation Maude Abbott (1927) considered that diag nosis should not be impossible on the basis of the increased mobility of the heart its occasional hypertrophy without apparent cause and its frequent displacement to the left Displacement to the left without displacement of the trachea can rightly be considered as a *diagnostic* pointer

Cert un complications are of importance in that they may lead to death Herniation of the heart may occur through the defect and strangulation ensue Such cases are those of Boxall (1887) in a woman who died three days after delivery with symptoms thought to be due to pulmonary embolism and the case of Sunderland and Wright Smith where pressure on a coronary artery by the firm edge of a defect was the cause of death in a child of two years Another risk is exposure of the heart to pulmonary infection with pleuropericarditis a cause of death in about a quarter of the cases A part of the liver may herniate into the pericardial sac through a diaphragmatic hernia (Gross 1946) Although rare such cases are of importance because of the possibility of surgical cure

PROGNOSIS *Southworth and Stevenson (1938) concluded that the* anomaly had no appreciable influence on life expectancy and found that ages at death ranged from birth to 75 years with a mean when infants under one year are excluded of 43 9 years Abbott gives 45 years as the average age at death

(2) DIVERTICULUM OF THE PERICARDIUM

HISTORICAL The first case appears to have been that of Hart (1837) who showed a specimen with a diverticulum in the anterior mediasti mum which communicated with the pericardial cavity Hird (1848) displayed to the Westminster Medical Society a diverticulum of the right side of the pericardium Peacock (1866) mentions Bird s case and another in the museum of St Thomas s Hospital

This condition is even rarer than a pericardial defect The lesion *consists of a protrusion of the parietal pericardium* so that a cyst like structure is formed This may become sealed off and even increase in size It may be associated with inflammatory disease of the pericardium and a rise in intrapericardial pressure predisposes to the condition Some cases are however developmental but in older patients where the possibility of inflammatory disease cannot be excluded the unreserved acceptance of a developmental origin is often impossible

Diverticula may arise from any part of the parietal pericardium but they are most common on the right side The diverticulum may be quite sealed off or may communicate with the pericardial cavity

by means of an orifice of varying size or even by a long tubular neck

There are no symptoms in the great majority of cases but in the case of Cushing (1937) a swelling was noted on the front of the chest Most often the condition is found when X ray examination of the chest is undertaken for pulmonary or cardiac disease It is thus of importance because it enters into the differential diagnosis of aneurysm or *mediastinal tumour*

Radiological examination discloses a rounded tumour intimately associated with the heart border and pulsation is present This can be clearly demonstrated with the radiokymograph and other radiological methods (Ernst 1935 Kernbock and Weiss 1934 Arrilaga Donovan and Taquini 1938) Cushing (1937) injected air into the diverticulum and this entered the pericardium

Unless there are pressure symptoms there is no obvious need for surgical extirpation

(3) ECTOPIA CORDIS Ectopia cordis is considered here owing to the frequency with which it is allied to a defect of the pericardium Ectopia cordis refers to that state where the heart lies completely or partially outside the thoracic cavity either on the surface or in the abdominal cavity The condition is relatively rare and Kellet (1936) found nineteen cases in the literature This is a low figure for Roth (1939) was able to collect 116 cases and Byron (1948) 141 cases The anomaly is so striking that few cases are likely to have remained unreported

Several types of ectopia may be recognized dependent upon the position of the heart In the *cervical* type the heart lies in the neck above the suprasternal notch and the sternum may or may not be fissured In the *pectoral* type the heart lies on the front of the chest and the sternum is fissured or defective or may be practically absent In a few cases according to Wedd (1929) the sternum may be normal except for a foramen through which pass the great vessels to and from the heart There is no covering by skin or other tissue In the *abdominal* type the heart passes through a defect in the diaphragm to reach the abdominal cavity It may remain entirely intra abdominal or it may appear on the surface of the body below the xiphoid cartilage and above the umbilicus Other types have been recognized and are combinations of the above

In the early stages of development the heart is subpharyngeal and in relation to the mandibular arch until about the fifth week Subsequently it becomes thoracic and the commonest type of ectopia is then related to defective development of the sternum Whether the defect of the sternum is a primary event and the ectopia a secondary phenomenon or whether the reverse is the case and adhesions between the heart and amnion cause ectopia are matters for speculation The longitudinal axis of the heart is generally abnormal and frequently the

apex points towards the chin and the base towards the abdomen so that the normal anterior surface of the heart comes in contact with the chest wall The pericardium is generally defective and frequently absent In the cervical heart the pericardium is always absent In some cases of pectoral type the heart partially issues through a defect of the sternum and has the appearance of a pulsatile hernia covered with skin or may even be without any covering Where the heart is without covering inflammatory changes rapidly set in and lead to speedy death Other cardiac anomalies may be associated as well as bodily deformity The tetralogy of Fallot was present in Byron s case The male sex is most frequently affected Death generally occurs early but a case of Foy (1910) where there was only a small sternal fissure lived to the age of 37 before dying of cardiac failure

Attempts at surgical treatment have so far been without success Cutler (1925) found no room in a chest where the tissues had become accommodated to the abnormal state and could only cover the heart with skin flaps under too great a tension Byron s (1948) attempt was unsuccessful Probably the only surgical approach would be an attempt to cover the heart with a pedicle graft

Rhabdomyomata

Fifty cases of this condition have been reported and have been summarized by Labate (1939) The rhabdomyomata are primary tumours of the heart Superficially their structure may resemble the Purkinje tissues but there is no connection with them The tumour histologically consists of large embryonic muscle fibres of greater diameter than in the normal heart muscle fibre and large vacuoles are present in the individual fibres Glycogen granules may be demon strated in the vacuoles by appropriate methods Striation may be visible at the margin of the fibres The types of rhabdomyomata are (1) solitary (2) multiple (3) diffuse The multiple type is often associ ated with tuberose sclerosis and adenoma sebaceum (Yater 1931 O Flynn and Mackay 1937 Labate 1939) The diffuse type where all of the heart muscle may be replaced by rhabdomyoma fibres is identical with the glycogen disease of von Gierke which furnishes many examples of the so called idiopathic hypertrophy of the heart

Although rhabdomyomata occur most commonly in young people their occurrence in the seventh decade has been observed by Bradley and Maxwell (1928) The naked eye appearance is of single or multiple rounded masses lying in the ventricular wall and projecting either from the external surface of the heart or into one of its cavities A heart cavity may be almost completely filled Reeves and Michael (1936) describe rupture of the heart at the site of the tumour In general the clinical effects of a rhabdomyoma depend upon its position and if it obstructs a valvular orifice death may be early

Congenital Idiopathic Hypertrophy of the Heart

The first case was reported by Simmonds in 1899 and since then about eighty similar cases have been described mainly on the basis of an enlarged heart occurring in infancy The work of Stoloff (1928) and Kugel and Stoloff (1933) and later Kugel (1939) has led to the elimination of more than two thirds of the cases originally described in this group

The term idiopathic hypertrophy of the heart was at first applied to a massive enlargement of the heart occurring in infants and young children Such a definition implies that there should be no recognizable etiology if the term idiopathic is to be retained The work of Kugel and Stoloff by analysis of fifty two cases revealed that there were only fifteen cases to which the term idiopathic might be applied The remaining cases all presented gross anatomical malformation capable of producing hypertrophy or microscopical evidence of inflammation and degeneration and thus are properly excluded from this group It is not improbable that these numbers will be further reduced when cases presenting a large heart are subject to detailed and critical examination

Kugel s (1939) classification of the causes of enlargement of the heart in infants and young children is of interest and value He lists the following conditions

(1) *Congenital defects*
- (*a*) Heart
- (*b*) Coronary arteries
- (*c*) Aorta and pulmonary artery

(2) *Infections*
- Unknown etiology
  - (*a*) Rheumatic fever
  - (*b*) Fiedler s myocarditis
  - (*c*) Periarteritis nodosa
- Known etiology
  - (*a*) Diphtheria
  - (*b*) Scarlet fever
  - (*c*) Subacute bacterial endocarditis with valve defect
  - (*d*) Syphilis

(3) *Anaemias*

(4) Syndrome of non suppurative myocardial degeneration with dilatation and hypertrophy

(5) *Metabolic*
- (*a*) Avitaminosis
- (*b*) Thyroid deficiency
- (*c*) Glycogen disease

(6) *Hypertension*

In greater circulation	(*a*) Essential (*b*) Adrenal tumours (*c*) Secondary to renal lesions
In lesser circulation	(*a*) Pulmonary lesions (*b*) Kyphoscoliosis

(7) *Tumours of heart*

(8) *Unclassified*

To this formidable list might be added the cardiac hypertrophy of the infants of prediabetic mothers described by Miller (1945) Certain of the above mentioned causes of hypertrophy of the heart in infancy are congenital in that they exist from birth Pompe (1933) reported a case of massive enlargement of the heart in a female child of seven months Microscopical examination of the heart showed fibres which were enlarged and vacuolated and appropriate staining showed these vacuoles to be filled with glycogen Sections of other organs revealed the same condition A similar case was described by von Gierke (1929) and he named the condition hepatonephromegalia glycogenitica more frequently known as von Gierke s disease Schminke (1922) described the heart of an infant in which were present hypertrophy of its walls and microscopical changes similar to those observed by Pompe He noted the vacuoles but did not stain for glycogen He assumed that the fibres were embryonic and as they were increased in numbers he thought the condition to be a neoplastic one and a type of rhabdo myoma The cases of Steiner and Bogin (1930) and Sprague Bland and White (1931) are also similar to that of Pompe although staining for glycogen was not undertaken Confirmed cases of glycogen disease are those of Deelman (1931) Putscher (1932) Antopol (1933) and Ellis (1935) A personal case presented itself as a failure to gain weight with feeding difficulties at the age of six months The child developed pneumonia of the right upper lobe at the age of seven months and X ray examination revealed a hitherto unsuspected enlarged heart Autopsy and microscopical examination revealed glycogen disease with involvement of the heart and liver (fig 13)

Other conditions which may give rise to hypertrophy of the heart are those dependent upon anatomical abnormality The most important type is an anomalous origin of the left coronary from the pulmonary artery Bland White and Garland (1933) and others have shown that inadequate nourishment of the heart muscle as a result of this anomaly leads to myocardial degeneration replacement fibrosis and later even aneurysmal dilatation of the heart wall In addition to the local changes in the territory of the abnormal coronary artery there is a well marked hypertrophy of the heart as a whole which in all probability represents

a compensatory mechanism This type may give the T wave changes in the electrocardiogram that are associated with coronary disease An unusual type of hypertrophy is furnished by those rare cyanotic cases where both pulmonary veins enter the coronary sinus In another group of cases, similar to that of Cathala (1926) hypertrophy was attributed to hypoplasia of the chromaffin system placing the cases in the cadre *of the myocardie of Laubry where failure may suddenly take place* in the absence of any valvular lesion and the microscopical appearances

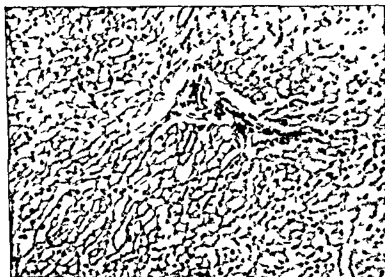


FIG 13 Heart muscle in von Gierke s disease Marked vacuolation of muscle fibres

are those of pure hypertrophy Avitaminosis of beri beri type may be the background of other cases Rosen s (1943) case revealed at autopsy the stigmata of congenital syphilis There was sclerosis of the pulmonary vessels as a cause of the cardiomegaly This case suggests the advis ability of serological tests in these individuals for treatment might well have had beneficial results

A new comer to this field is the cardiac hypertrophy and extra medullary erythropoiesis in new born infants of diabetic or pre-diabetic mothers described by Miller (1945) Recognition of these cases presents difficulties Increased birth weight normoblastaemia and cardiac hypertrophy are pointers to diagnosis Cyanotic attacks dyspnoea and tachypnoea are suggestive symptoms The heart returns to a normal size within the first or second month and cardiac and

respiratory symptoms do not persist beyond the first few days in contrast with hypertrophy due to other causes

Despite the rapidly narrowing field of idiopathic hypertrophy there still remains a certain number of cases where microscopical examination has shown no abnormality other than an increase in the number of fibres Examples are those of Moncrieff (1929) and Debré and Busson (1931) *A familial tendency or congenital weakness of the germ plasm* has been discussed by Sprague Bland and White (1931)

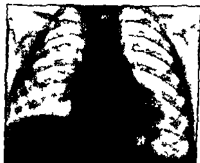


FIG 14 Radiograph of chest of seven months female infant showing enlargement of the heart Autopsy showed glycogen disease

CLINICAL PICTURE The clinical picture will to some extent depend upon the underlying pathology The usual history is that of a child born at term and is apparently normal for some weeks or months before symptoms appear Occasionally symptoms may be apparent from the first day of life The commonest symptoms are dyspnoea and cyanosis although an abnormal pallor has been noted in some cases (Debre and Busson 1931) Dyspnoea may be paroxysmal or continuous and cyanosis may only be evident during a paroxysm Stridor may be present and the lower intercostal spaces may be sucked in with inspiratory efforts Cough is often present and may resemble whooping cough (Stoloff 1928) Examination of the chest shows impairment of resonance on the left side with a diminished air entry into the left base due to compression of the left bronchus In Stephenson s case reported by Abbott there was hoarseness from pressure on the left recurrent laryngeal nerve atelectasis and also dysphagia and obstruction to the passage of a feeding tube

A rapid heart rate is generally present with sounds of poor quality Murmurs are often absent although a soft blowing systolic murmur has been described A gallop rhythm may be present

Sometimes clinical examination of the chest is negative and a routine X ray examination may furnish the first evidence of the massive enlarge

ment of the heart The insidious onset and possibly vague early symptoms conspire to make this one of the most infrequently clinically recognized abnormalities Anomalous origin of a coronary artery is more fully discussed on a later page

RADIOLOGY The heart is massively enlarged to the left and right and vertically On the left side the heart may appear almost to fill the left chest The discovery of enormous hypertrophy in an infant showing symptoms of dyspnoea and cyanosis is adequate diagnosis It at once

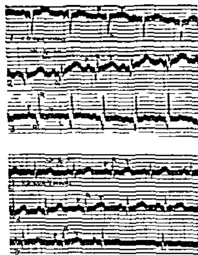


FIG 15 Electrocardiogram from same case as fig 11 at eleven days and seven months Note the physiological change in axis deviation

suggests that children with these symptoms should always be examined radiologically for by these means evidence is sometimes obtained that may be clinically lacking A case of glycogen disease in an infant is shown in fig 14

ELECTROCARDIOGRAM The electrocardiogram has been taken in a few cases In all cases reported there has been a tachycardia of sinus origin Stoloff s case showed low voltage and shallow T waves There was normal axis deviation in the case of Sprague Bland and White Inversion of the T waves suggests an abnormal origin of the coronary arteries (Bland White and Garland) A personal case of glycogen disease showed right axis deviation at eleven days and normal axis deviation just before death at seven months (fig 15)

COURSE AND PROGNOSIS This naturally depends upon the basic course of enlargement Once symptoms have appeared the general

course is downwards and death usually results from pulmonary infection and broncho penumonia Some cases have died of failure with oedema Death usually occurs within the first year although life may be prolonged to the age of four

Cardiac Aneurysm

A few cases of cardiac aneurysm have been attributed to congenital developmental defects The condition is mentioned by Abbott (1927) in reference to a 2½ month old child with congenital syphilis with an aneurysm at the apex of the left ventricle In a second case there was a pulsating mass of tissue passing from the apex of the left ventricle through a defect of the diaphragm to form part of an umbilical hernia Ungerleider and Gruber (1945) mention an aneurysm of the left ventricle in a girl of 15 thought to be due to abnormality of development of the muscle bundles Cardiac aneurysm may also occur in association with abnormal origin of the left coronary artery from the pulmonary artery as for example in the cases of Sanes and Kenny (1934) and Linck (1936) Bland White and Garland (1933) collected eight similar cases from the literature It should be emphasized that in late childhood and adolescence it is possible for an aneurysm of the heart to result from necrosis of cardiac muscle due to rheumatic carditis (Parkinson Bedford and Thomson 1938 Burn Hollander and Crawford 1943)

The abnormality will generally be recognized by its radiological picture of a bulge on the left cardiac border giving the heart a square appearance with a shelf like superior border Paradoxical pulsation may be detected on screen examination the affected area expanding when other parts of the heart contract in systole The reported cases have in some instances been associated with abnormal electrocardio grams These changes are not specific and are similar in type to those found in cases where a left coronary artery arises from the pulmonary artery

CHAPTER V

AORTIC AND SUBAORTIC STENOSIS

Aortic Stenosis

Stenosis of the aorta may occur at the valve at a point distal to the insertion of the ductus arteriosus (coarctation) and rarely near the bifurcation of the aorta A further type of stenosis occurs proximal to the valve in the aortic conus A generalized narrowing of the aorta associated with hypoplasia and increased elasticity of its walls is termed aortic hypoplasia While this latter may be a primary anomaly it is most often associated with other congenital defects

The types of aortic stenosis may be classified as follows

(1) Valvular stenosis
 (*a*) Developmental
 (*b*) Inflammatory
 (*c*) Pure aortic stenosis (Gallavardin)
(2) Aortic atresia
 (*a*) Inflammatory
 (*b*) Developmental
(3) Subaortic stenosis
 (*a*) Developmental
 (*b*) Due to hypertrophy of the tissues of the aortic conus
(4) Stenosis of the aortic isthmus
(5) Stenosis of the aorta distal to the isthmus
(6) Hypoplasia of the aorta

Aortic atresia isthmus stenosis and stenosis distal to the isthmus are considered under anomalies of the aortic arches (page 56)

Valvular Aortic Stenosis

Pathogenesis The aortic valves are evolved from the proximal ends of the distal bulbar swellings This event occurs in the seventh week at about the time that the septa are closing and in practice is comparatively rare to find defects of the interventricular septum associated with this type of stenosis A second and commoner type is due to an endocarditis of the aortic valves after the septa have closed this condition being the homologue of pulmonary stenosis with a closed septum Farber and Hubbard (1933) have collected five such cases in which a foetal endomyocarditis appeared to be responsible for a valvular aortic stenosis A special type of valvular aortic stenosis described by Gallavardin (1936) and others as pure aortic stenosis and presumably due

to an infective process of early origin and unknown etiology is discussed elsewhere (page 49)

ANATOMY The valve cusps are thickened and may be fused together to form a diaphragm with a central opening Variations in size and number of the cusps may occur and sclerosis in these abnormal cusps leads to obstruction Calcification may occur in the thickened cusps and there is the same liability to infective endocarditis as exists in other forms of congenital valvular anomaly There is some degree of left ventricular hypertrophy proportional to the degree of valvular stenosis Distal to the obstruction the aorta may be dilated The explanation of this is obscure but in all probability the dilation is due to a hypoplasic aortic wall As indicated above the possible origin by infective process after the septa have closed makes associated cardiac abnormalities rare and the presence of an interventricular septal defect is most uncommon On the other hand other defects of the aorta or aortic conus may be coincident Subaortic stenosis coexisted with valvular stenosis in the cases of Smart (1904) and Shennan (1908) with disease of the mitral valve in those of Kockel (1908) and Farber and Hubbard (1933) Some of these cases must support the concept of an inflammatory origin of subaortic stenosis Hypoplasia of the aorta is frequently present and is a comparable finding to the aortic hypo plasia of mitral stenosis of standing

CLINICAL PICTURE The subjects of aortic stenosis may be of small statute and the occurrence of dwarfism is well recognized in this condition Pallor is common and has been attributed to arterial depletion However in those cases where stenosis is slight these signs may be minimal Symptoms are relatively few and the abnormality may be entirely latent and only discovered at a routine physical examination at school entrance or for military service

The physical signs are striking The heart is slightly enlarged and the apex beat forcible Usually there is a systolic thrill at the base and to the right of the sternum The thrill may be felt in the carotids Absence of a thrill need not invalidate diagnosis A harsh systolic murmur may be heard over the praecordium with maximum intensity in the second right space Localization of thrill and murmur at this site is characteristic of aortic obstruction The murmur is transmitted along the subclavians to the axilla and may be heard in the neck and back The second sound tends to be diminished or absent A pulsus tardus is present and the blood pressure is low Correspondingly the pulse is small and may be difficult to feel The contrast between a small pulse and rather forcible apex beat is a very suggestive and valuable sign of aortic stenosis In a few cases a diastolic murmur of aortic in competence may be heard along the left sternal margin Despite these signs diagnosis may at times be difficult Evans and Lewes (1945) reported a carotid shudder in those cases where aortic incompetence

accompanies aortic stenosis Detectable over the carotids at the height of systole there is a momentary visible quiver or vibration This sign may be recorded with the polygraph and occurs at the height of the up stroke Polygraph examination is also of use in those cases where aortic stenosis is uncomplicated by incompetence and identical tracings are obtained This may cast doubt upon the specificity of the carotid shudder although as a visible phenomenon it may occur solely in the combined lesion Grishman Steinberg and Sussman (1947) find a characteristic arterial pulse with slow rise anacrotic notch systolic vibrations and systolic plateau This however occurs in both aortic and subaortic stenosis and in aortic stenosis with incompetence

RADIOLOGY The general appearance of the cardiac silhouette corresponds closely with that found in other types of aortic disease There may be slight or moderate hypertrophy of the left ventricle The ascending aorta is often dilated and the aortic knuckle small and inconspicuous

ELECTROCARDIOGRAM The electrocardiogram shows normal or left axis deviation Evans (1948) states that a right axis deviation is sometimes present

COURSE AND PROGNOSIS Apart from the special risks of an infective endocarditis involving the valve cusps the subject is exposed to the same risks as other types of aortic stenosis Syncopal attacks may occur on exertion and in older subjects there may be pain of anginal type provoked by effort There is a distinct liability to sudden death particularly in cases with calcified aortic cusps There is some tendency to the development of pulmonary tuberculosis

The lesion is frequently well tolerated and no symptoms may occur to draw attention to the heart The discovery of a heart lesion may be purely accidental Although the condition appears to be compatible in many cases with a reasonably normal life it is wise to restrict activities in relation to exercise or tasks that involve considerable effort

The average age at death in Abbott s (1931) series was 3 75 years with an extreme of 24 years Undoubtedly cases survive beyond this age

PURE AORTIC STENOSIS

(Syn Retrécissement aortique pur non rheumatismal – Gallavardin)

A brief account of this lesion is given because it is held by some to be of congenital etiology

DEFINITION Stenosis of the aortic valve arising in infancy as the result of an unknown infection (Gallavardin)

The greatest number of these cases are in males while in the rheumatic infection the chief incidence of aortic valvulitis is in the female Although nothing is definitely known about the etiology of these cases it is tolerably certain that they bear no relation to either syphilis tuberculosis or rheumatism The pathological process is a

sclerosing endocarditis of the aortic valve This may arise in foetal life in a similar manner to the foetal endocarditis that determines a pulmonary stenosis with a closed septum It may possibly arise in early post natal life If it is accepted that the lesion predominantly arises in foetal life it must then be classified as a congenital abnormality The designation pure relates to the absence of aortic incompetence Gallavardin contends that pure *aortic stenosis is a guarantee of its* non rheumatic etiology as rheumatic aortic stenosis is invariably accompanied by aortic incompetence The post mortem features are those of sclerosis and scarring of the aortic cusps

CLINICAL PICTURE Symptoms are slight or entirely absent the discovery of a heart lesion being often purely fortuitous The symptomatology depends upon the degree of stenosis present In severe cases there may be infantilism of the Lorain type Syncope and angina of effort may occur Infective endocarditis may involve the stenotic valve

The physical signs are those of other types of aortic stenosis A systolic murmur and thrill maximum in the second right space with *conduction of the murmur into the vessels of the neck is characteristic* The pulse is small and the blood pressure low In 28 per cent of Gallavardin s cases a diastolic murmur was present but was unaccompanied by the major peripheral signs of aortic incompetence

RADIOLOGY The X ray shows lowering of the apex and rounding of the left ventricle proportional to the degree of stenosis present The ascending aorta is dilated

ELECTROCARDIOGRAM The electrocardiogram shows normal or left axis deviation

COURSE AND PROGNOSIS The lesion is well tolerated in young subjects and may not give rise to any symptoms The lesion is compatible with *long life (Gallavardin)*

To the British observer it would appear to be academic to segregate this group of cases from what are generally accepted as cases of true congenital aortic stenosis

Subaortic Stenosis (Syn Infundibular subvalvular stenosis)

Subaortic stenosis consists of a narrowing of that part of the left ventricle situated immediately below the aortic cusps

ETIOLOGY AND PATHOGENESIS This abnormality is admirably explained on embryological grounds as its site is in that part of the left ventricle which is derived from the bulbus cordis the rest of the ventricle being derived from the primitive ventricle Normally the major portion of the bulbus cordis is incorporated in the right ventricle to form the conus of the pulmonary artery It largely disappears or atrophies in the left ventricle but a small portion contributes to the formation of the upper part of the interventricular septum *Failure of* this process of atrophy leads to subaortic stenosis Its homologue in the

right ventricle is conus stenosis of the pulmonary artery As the bulbus contributes to the formation of the upper part of the septum it follows that other abnormalities may be associated with subaortic stenosis These are aortic valvular stenosis (double aortic stenosis) bicuspid aortic valves defects of the interventricular septum and hypoplasia of the aorta Coarctation was present in the cases of Pilod and Hugonot (1926) and Stewart and Bellet (1934) Sometimes the stenosis may be so severe as to amount to atresia This is exceptional and when it occurs a defect of the interventricular septum hypertrophy of the right ventricle and a patent ductus arteriosus are present Pure muscular hypertrophy of the upper part of the interventricular septum has been described as a congenital condition

In addition to subaortic stenosis of purely developmental origin there are some acquired types which should be considered as these may be attributed to foetal endocarditis Thus there are those cases reported by French authors of subaortic stenosis with mitral stenosis Two origins of these cases are possible Either extension of a rheumatic mitral valvulitis to the subaortic endocardium so that a subaortic shelf is formed (Vulpian 1868 Liouville 1869) or a thickened aortic cusp of the mitral valve may extend into the ventricular cavity below the aortic valve as in the case of Lemierre and Bernard (1925) In either case calcification may occur Muscular hypertrophy of the upper part of the septum sufficient to narrow the aortic conus is seen in advanced hypertension and in lesions causing concentric hypertrophy of the ventricle

ANATOMY The site of the lesion is five to ten millimetres below the aortic cusps Usually it takes the form of a collar of fibrous tissue encircling the aortic conus and continued on to the anterior cusp of the mitral valve (fig 16) The case of Christian (1933) showed a membranous structure with a small opening Thus a diaphragm may be formed or else the lesion may be simply a crescentic fold of fibrous tissue or of endocardial thickening Microscopical examination of the lesion has shown that it largely consists of elastic tissue continuous with the subendocardial elastic tissue of the heart (Wigglesworth 1936). This constitutes very strong evidence of a congenital origin and renders doubtful any possible inflammatory or mechanical origin Whatever form the lesion may take it is liable to become the seat of an infective endocarditis or to become calcified

Occasionally there coexists a stenosis of the aortic valve the condition then being referred to as double aortic stenosis (Smart 1904 Shennan 1908) With the onset of infective endocarditis the aortic cusps are frequently involved as in Christian s case and in a lesser number of cases the subaortic lesion Hypoplasia of the aortic wall is commonly present and in life there may be dilatation of the aorta distal to the lesion

CLINICAL PICTURE Subaortic stenosis of itself does not as a rule offer any very serious obstruction to the egress of blood from the left ventricle Except in the severest deformities the peripheral signs of aortic stenosis may be lacking Cases do not show the same tendency to dwarfism or under development that is presented by valvular stenosis

The lesion may be entirely latent and only discovered at a routine school medical examination or when life assurance is contemplated In later years the significance of the finding may be overlooked and the



FIG 16 Subaortic stenosis in a young female who died suddenly whilst cycling A shelf of fibrous tissue stretches across the aortic conus M severed cusp of mitral valve A aorta LA left auricle

signs interpreted as those of acquired disease In the severe degrees of subaortic stenosis there may be shortness of breath on exertion and children with the lesion may not be as energetic as their fellows In some of the recorded cases there has been precordial pain on exertion and in a few cases convulsions have occurred these latter being attributed to transitory cerebral anaemia Such symptoms would appear to be far more likely in the valvular type of aortic stenosis

The heart may be normal in size or slightly enlarged enlargement being dependent upon the severity of the lesion and upon the presence or absence of other defects A systolic thrill may be felt diffusely over the praecordium but is usually localized to the base of the heart It may be felt over the supraclavicular fossae and in the episternal notch The thrill is of maximum intensity in the first and second right spaces Muller (1924) suggested that the position of the thrill varied with the position of the patient and that when recumbent the thrill was defi nitely most perceptible at the base In the upright position the thrill was more often apparent at the apex The apex beat is forcible A loud harsh systolic murmur occupying the whole of the systole and widely

heard over the heart is maximum in the *second right* space The bruit is characteristically transmitted along the great vessels Such conduction of a murmur comparable to that observed in valvular stenosis is very suggestive of a lesion of the aortic tract The aortic second sound is normal and unaltered

An experience of forty three cases for the most part in children and young adults suggests that in about a third of the cases despite the obvious physical signs of aortic stenosis the pulse appears to be normal



FIG 17 Male aged 14 presenting typical signs of subaortic stenosis The aorta is further to the right than normal and the aortic knuckle is small

and the blood pressure not necessarily low This view is shared by Young (1944) who recognized twelve such cases in soldiers between the ages of 19 and 30 who were accustomed to severe exertion and symptomless

More often the pulse rate is small and the blood pressure low The polygraphic examination reveals a pulse of similar characters to that of aortic valvular stenosis mainly a slow rise anacrotic notch systolic vibrations and a plateau

RADIOLOGY The radiological picture may be typical of that of aortic stenosis from any cause The general picture is of increased convexity of the left ventricle a small aortic knob and a dilated ascending aorta (fig 17) There is no change in the pulmonary arc in uncomplicated cases

ELECTROCARDIOGRAM The electrocardiogram shows normal or left axis deviation A *normal* axis in no way excludes subaortic or indeed aortic stenosis (fig 18)

DIAGNOSIS Diagnosis from other forms of aortic obstructive lesions may be a matter of considerable difficulty It may be said that there is no single pathognomic sign of subaortic stenosis which serves to differentiate it from stenosis of the valvular type

Grisham Steinberg and Sussman reviewing twenty three of their cases could find no distinguishing sign between the two conditions A diastolic murmur is perhaps more sugge tive of aortic valvular stenosis but Walsh Connerty and White (1943) have found a diastolic murmur

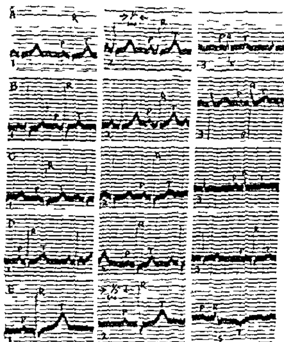


FIG 18 The electrocardiogram in five children with signs of subaortic stenosis

in a case of subaortic stenosis with a bicuspid aortic valve Points in favour of a subaortic stenosis are knowledge of a heart lesion since infancy an absence of any considerable enlargement of the heart marked contrast between the physical signs of aortic obstruction and the normal well being and development of the subject and a normal unaltered aortic second sound Absence of a rheumatic history in a subject below the age of 20 together with the absence of other valvular lesions or gross enlargement of the heart should also favour a con genital etiology Perhaps the newer method of angiocardiography will

ultimately furnish the most reliable information and permit certain diagnosis between the two conditions

COURSE AND PROGNOSIS Subaortic stenosis is a relatively benign lesion many cases being entirely symptomless and thus overlooked The abnormality may be compatible with a normal life span the maximum age at death in Abbott s series being 58 A patient of Stein berg (1933) lived to the age of 77 Grishman *et al* (1947) and Young (1944) consider the prognosis to be rather poor The chief danger is infective endocarditis which can occur at any time but it is more frequent in the second and third decades Syncopal attacks and sudden death may occur with no lesion other than the stenosis There is some evidence that there is an increased liability to pulmonary tuberculosis

CONGENITAL AORTIC INCOMPETENCE The existence of aortic incompetence as a congenital abnormality is extremely rare and White (1944) is of the opinion that no such case has so far been reported There are however certain congenital conditions in which aortic incompetence may occur Complete absence of the aortic valve is mentioned by Abbott (1927) Aortic incompetence has been found with bicuspid aortic valves *but infrequently unless there has been added* infection of the valve It also occurs with fenestrated valves Froment Bertoye and Perreau (1947) reported an incompetent quadricuspid aortic valve in a man of 65 Incompetence may coexist with congenital aortic stenosis and occasionally in subaortic stenosis An isolated interventricular septal defect may occasionally develop deformity of an aortic cusp with resulting incompetence In the cyanotic group the large dextroposed aorta of the Fallot has been occasionally accom panied by incompetent valves Likewise Taussig and Semans (1940) have noted incompetence in an Eisenmenger complex with coarctation and it can occur with congenital aortic aneurysms It appears that aortic incompetence is probably exceedingly rare as an isolated con genital abnormality and that when present it is most often accompanied by other congenital anomalies of the heart and aorta which have been subjected to either strain or infection

CHAPTER VI

CONGENITAL STENOSIS AND ATRESIA OF THE AORTIC ARCH

The various anatomical types of stenosis and atresia of the aortic arc (fig 19) have been classified by Evans (1933) as follows

(1) Stenosis of the arch with hypoplasia of the proximal aorta Pater ductus arteriosus
(2) Stenosis of the arch with hypertrophy of the proximal aorta Ductus arteriosus closed
(3) Congenital atresia of the distal part of the arch Hypertrophy o the proximal aorta Ductus arteriosus closed
(4) Interruption of the aortic arch in its distal portion Hypoplasia of the proximal aorta Ductus arteriosus widely patent and continuous with the descending aorta
(5) Congenital atresia of the proximal part of the arch Patent ductus arteriosus
(6) Absence of the ascending aorta Ductus arteriosus patent

To these may be added for the sake of completeness

(7) Stenosis of the aorta distal to the isthmus

Types (1) (2) and (3) correspond in broad terms with those cases generally classified as coarctation of the aorta and their distinction depends upon the degree of stenosis and status of the ductus arteriosus Bramwell (1947) suggests a simplification of this classification by dividing the six types into three groups Group 1 would comprise Evans s type (2) and (3) in both of which the ductus is closed Group 2 includes Evans s type (1) and (4) both of which are characterized by a hypoplastic proximal aorta and a patent ductus arteriosus Types (5) and (6) form a third group where the patent ductus arteriosus is the only channel between the heart and aorta the ascending aorta being occluded or absent From the point of view of prognosis the expectation of life is poor in group 2 and in group 3 cases never survive infancy Expressed in other terms the members of groups 2 and 3 have the infantile type of coarctation which is often accompanied by other grave errors of development whereas in group 1 which comprises the adult type of coarctation grave developmental cardiac anomalies are uncommon From the embryological point of view group 1 is con cerned with abnormality or arrest of development in the region of the junction of the fourth and sixth primitive arches The remaining types result from abnormal development of the truncus arteriosus and con

sequently other abnormalities such as transposition of the vessels pulmonary atresia or bilocular or trilocular heart may be present

On clinical grounds only the adult type of coarctation is important and capable of positive recognition during life

STENOSIS OF THE AORTIC ISTHMUS COARCTATION OF THE AORTA

DEFINITION Stenosis or obliteration of the arch of the aorta at a point distal to the origin of the left subclavian artery and at or adjacent to the site of insertion of the ductus arteriosus

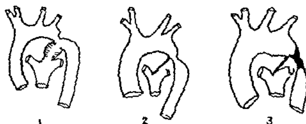


FIG 19 Types of coarctation of the aorta (After Evans) 1 Narrowing of the isthmus and ductus arteriosus patent 2 Narrowing of the isthmus ductus closed 3 Obliteration of the isthmus ductus closed

HISTORICAL NOTE The first hint of a case of coarctation comes from Morgagni (1760) who discussing a post morten observation stated that the aorta was contracted to an amazing narrowness near the heart Paris (1791) described the collateral circulation in the first really authenticated case of coarctation Meckel (1827) furnished his well known plate of the collateral circulation in the thorax This was the first plate to depict notching of the under surfaces of the ribs and it also shows notching of the upper surfaces of the third and fourth right ribs Jordan (1830) in his description of a case a body that had been raised for the purposes of dissection states that the ribs had become deeply absorbed and sulcated in some parts by the pressure of the branches of this artery Legrand (1835) appears to have made the first clinical diagnosis of aortic obstruction during life on the basis of a collateral circulation and reduced femoral pulsation Mercier (1838) first employed the term coarctation Craigie was able to collect nine cases in 1841 and added one of his own Oppolzer (1848) is considered by some to have made the first clinical diagnosis of coarctation at its usual site Scheele (1870) effected simultaneous tracings of the radial and femoral pulses Potain (1892) is stated by Flexner (1930) to have noted hypertension in the upper extremities Abbott (1928) analysed 200 cases from the literature The important radiological sign of rib notching was first noted and emphasized by Railsbach and Dock (1929)

The first account of the surgery of coarctation is that of Crafoord and Nylin (1945)

ANATOMY The classification of Bonnet (1903) is often adhered to and is generally accepted Bonnet divided coarctation into two types the infantile and the adult The infantile consists of a diffuse narrowing of the aortic isthmus This is really a persistence of the condition of the aorta in foetal life and Theremin (1895) stated that such a narrowing might be found in 80 per cent of infants during the first three months

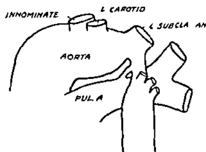


FIG 20 Diagram of coarctation of the aorta

after birth In the adult type there is an abrupt constriction at or near the level of the insertion of ductus arteriosus and he inclined to the view that this was an acquired lesion The adult type is unquestionably the more important condition and is the anomaly that is likely to be encountered and recognized Narrowing or obliteration of the arch does not always occur precisely at this site It may occur just distal to the left carotid artery and proximal to the left subclavian artery These abnormal sites of stricture give rise to differences in the clinical picture and will be discussed below In the adult type of coarctation the ductus is most often closed If it remains patent it may provide a channel for blood to reach the aorta distal to the coarctation and thus remove the necessity for a well developed collateral circulation Abbott s (1928) series of 200 collected cases lists eighteen in which the ductus was patent

The external aspect of the aorta may show sudden constriction resembling an hour glass as if a ligature had been applied (fig 20) In other cases the aorta gradually tapers from the origin of the left sub clavian to the point of constriction and then it rather abruptly expands again the distal expansion being occasionally bulbous owing to the needs of the collateral circulation The interior of the aorta at the level of the lesion shows a thickening of the inner coats of the vessel so as to form a smooth diaphragm with a central opening of varying size down to complete obliteration Often the inner coat appears to be healthy but there may be atheromatous lesions and calcification at the site of the coarctation and just above Microscopically the aorta may

appear normal A diminution of elastic tissue with connective tissue replacement has been described at the site of stricture by Ascenzi (1947)

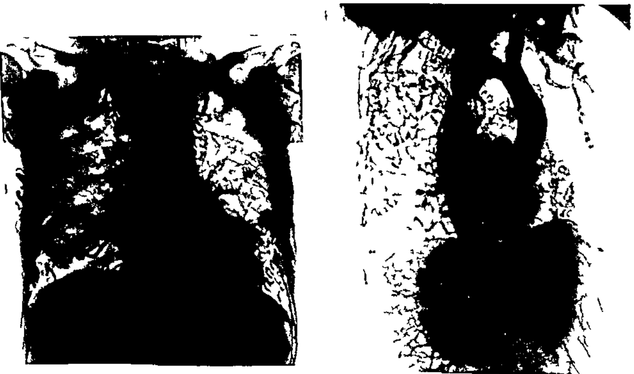
The aorta proximal to the obstruction may be thin walled and hypoplasic and in a small percentage of cases there may be a true developmental hypoplasia of the aorta which is narrower than normal The aorta is more commonly dilated and projects to the right The vessels of the arch are dilated and may have anomalous origins Atheromatous and degenerative changes in the aorta and vessels of the head and neck are common An atheromatous plaque may become the seat of a dissecting aneurysm with subsequent rupture Below the constriction the upper intercostals are enlarged and dilated as also the superior epigastric artery It is upon these vessels that there falls a major share in the maintenance of a collateral circulation to the lower parts of the body

Not infrequently there are associated cardiac anomalies Abbott (1928) has pointed out that grave cardiac defects may be found with coarctation of the infantile type whilst their association with the adult type is rare The commonest complicating anomaly is the bicuspid aortic valve present in 22 per cent of Abbott s 200 analysed cases and in over 50 per cent of the cases where rupture of the ascending aorta led to death Subaortic stenosis aortic stenosis of the valvular type and congenital aneurysms of the sinus of Valsalva have been noted Anomalies of the arteries of the aortic arch are frequently found and these are of interest as they perhaps throw some light on the genesis of the abnormality which in coarctation appears to arise at the point of junction of the fourth and sixth left arches It is reasonable to suppose that from the same cause irregularities may occur coincidently in the remaining vascular arches Thus amongst the vascular anomalies that have been described in coarctation are the origin of both carotids from a common trunk innominate and left carotid forming a common trunk abnormal origin of the right subclavian below the left and running behind the trachea and oesophagus suggesting a persistent fifth left arch New vessels may be developed from the aorta to assist in the formation of a collateral circulation

COLLATERAL CIRCULATION This is gradually developed in those cases where the obstruction is severe It is necessary in order that a circulation can be maintained below the level of the lesion With the development of such collateral channels the subject can often live comfortably provided there are no subsequent rheumatic or other infections and that the heart muscle is capable of supporting the strain that is placed upon it The occurrence of a collateral circulation was early recognised and was well figured by such early workers as Meckel (1827) and Jordan (1830)

The main paths of the collateral circulation are as follows

(1) Between the superior intercostal branches of the subclavians and the first intercostal within the chest



(*a*) Anteroposterior view (*b*) Oblique view

Fig 21 Injected arteries in a case of coarctation of the aorta Female aged 56 who developed hypostatic pneumonia subsequent to a fall (Mr Chesterman s case)

(2) Between the scapular branches of the subclavians and the upper aortic intercostals on the thoracic wall

(3) Between the internal mammary arteries and the epigastric branches of the external iliacs in the abdominal wall

These collateral pathways form tortuous and dilated arteries which may be visible and palpable on the back and abdominal wall (fig 21) They also erode the ribs a feature which was early recognized and an anatomical fact of supreme importance in radiological diagnosis

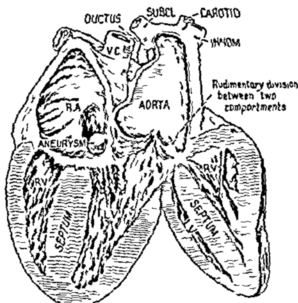


Fig 22 Coarctation of the aorta in female aged 30 who died of infective endocarditis involving the aortic cusps A mycotic aneurysm projects into the right auricle

Hypertrophy and dilatation of the heart especially of the left ventricle is often present and is the result of hypertension but is by no means invariable Unless there has been intercurrent infection such as rheumatism the valves are usually healthy and competent Even a bicuspid aortic valve may be perfectly competent Once however the factor of infection or degeneration is introduced and valvular deformity established a large heart is the rule There is a tendency for sclerotic changes to occur in the cusps which leads to deformity and results in incompetence Infective endocarditis occurs at the site of coarctation distal to the stricture or at the bicuspid valve It may also result in an arteritis of the ascending aorta In either case mycotic aneurysms or septic emboli may result (fig 22)

BICUSPID AORTIC VALVES A bicuspid aortic valve is the commonest associated lesion and occurred in 23 5 per cent of Abbott s series Reifenstein *et al* (1947) note this association in 42 per cent of their collected cases The importance of the bicuspid valve is in its liability to infective endocarditis and nearly three quarters of the patients with coarctation and infective endocarditis are found to have this valvular anomaly This is interesting in relation to the observation of Abbott (1932) who states that the incidence of bacterial endocarditis where a bicuspid aortic valve exists as a sole anomaly is about 40 per cent

The distinction between a congenital and an acquired bicuspid valve may present great difficulties and require microscopical examination There is some evidence to show that the bicuspid valve is prone to rheumatic involvement and Reifenstein found evidence of rheumatic valvulitis in two fifths of the cases with bicuspid valves Therefore it seems that the bicuspid valve may be prone to infective endocarditis either because it has been previously damaged by rheumatic valvulitis or because it is congenitally bicuspid

CONGENITAL ANEURYSMS The occurrence of congenital aneurysms of the cerebral vessels in association with coarctation has long been recognized In coarctation there may be a congenital weakness of the media (Abbott) and the existence of these congenitally defective areas has been confirmed by Symonds (1924) and Parkes Weber and Bode (1926) These may in some instances be the starting place of haemorrhage without actual aneurysm formation In coarctation there is a persistent hypertension and this may influence aneurysm formation in the defective areas Bleeding at first may be only slight and intermittent but ultimately it may be severe The post mortem examination of such an aneurysm may reveal by the varying age of the blood clot present that several spontaneous haemorrhages have taken place The clinical picture is that of a spontaneous subarachnoid haemorrhage The cases of Parkes Weber (1927) and of Woltman and Sheldon (1927) are illustrations of these coexisting lesions of coarctation and cerebral aneurysm Abbott on the basis of her analysis concludes that spontaneous cerebral haemorrhage occurring in coarctation before the end of the second decade in the absence of infection is due to rupture of a cerebral aneurysm

PATHOGENESIS It is an observation of fundamental importance that whilst the infantile type of coarctation is frequently observed before birth the adult type with severe constriction or complete obliteration of the aorta has never been found before or at birth Thus the adult type of coarctation has a post natal origin

Craigie (1841) enunciated and Skoda supported what is known as the Skodaic theory Craigie s view was that the obliterating process taking place in the ductus arteriosus extended to and involved the aorta causing its constriction Brunner (1898) suggested that the ductus

tissue was prolonged into the aorta at the insertion of the ductus arteriosus This view has never received any real anatomical confirma tion until recently, although the subject has been well studied by Klotz (1907) and others Pezzi and Agostini (1937) have however shown the penetration of the muscle tissue of the ductus into the wall of the aorta where it becomes continuous with the musculo-elastic layer It is also to be observed that the site of constriction may be at some distance from the insertion of the ductus and in some cases the aortic portion of the ductus remains open whilst the pulmonary end is closed yet constriction of the aorta has been complete In addition the pulmonary artery is never constricted in the process of involution of the ductus Such considerations as these latter do not entirely reconcile with a theory that must necessarily postulate the inclusion of ductus tissue in the wall of the aorta So also on the grounds of pure physiology the theory might be refuted for with the transition from the foetal to the post natal circulation the pressure in the aorta exceeds that in the ductus a circumstance unfavourable for a narrowing of the aorta

Previously to Craigie Reynaud (1828) has suggested that the abnor mality was a congenital one The isthmus of the aorta is derived from the fourth left arch The fifth left arch disappears and the sixth left arch which forms the ductus undergoes involution The site of coarctation is where these three primitive arches meet It seems possible that delay in involution of the fourth and sixth arches together with the pull exerted by the obliterating ductus on these embryonic tissues may be the cause of the occlusive process in the aorta

Ascenzi (1947) the most recent contributor to this problem found on microscopic examination of the site of coarctation that the middle elastic coat of the aorta was largely replaced by muscular tissue derived from the ductus arteriosus this fibro muscular tissue being most marked near the entrance of the ductus An excessive amount of this tissue tended to strangle the aorta at this site and prevent its development A deter mining factor was the traction exerted by the involuting ductus in the earliest days of life In further proof he cites the common presence of traction or torsion areas on the left posterior border of the aortic arch in infants and also mentions the rare cases of coarctation with traction aneurysms at the level of insertion of the ductus for example those of Laennec and Willigk (1853)

Clinical Picture Coarctation may be entirely latent and unsus p cted during life It occurs most frequently in males and in Abbot s (1928) 200 collected cases only a quarter were females There is no facile explanation of the increased frequency in the male but it has been advanced that the more sheltered existence of women exposes them to less physical strain than the male and a a result symptoms and complications are in abeyance and less likely to be detected Per sonal experience suggests that it may be more common in the female

than statistics indicate Lack of the awareness of the condition may easily be responsible for the few cases detected although hypertension is common enough in women A fruitful source of cases to the author has been the ante natal clinic from whence cases have been referred as hypertension without albuminuria It is not uncommon in children if the elementary precaution is taken of feeling the femoral artery as part of the routine examination In adult men it is often missed and cases may be found in the armed forces often engaged in the most strenuous occupations In sum our knowledge of its incidence must undergo revision with the increased impetus to its recognition furnished by the possibility of surgical treatment

Symptoms are uncommon in childhood and in the early age groups the finding of a case is largely a matter of careful routine examination often during some intercurrent illness It is important to critically review any case with a diagnosis of congenital heart disease or even of rheumatic heart disease especially in those cases where there is no clear cut history of rheumatic fever Common symptoms in childhood are a liability to epistaxis especially after exertion dyspnoea on exertion and very occasionally a subarachnoid haemorrhage

Most often symptoms develop in early adult life about the third decade at the period when strain is most likely to occur Early symptoms are of a banal type such as palpitation and dyspnoea Other and often more important symptoms are dependent upon the abnormalities of the circulation in coarctation and the adequacy of a collateral circulation perhaps determines the signs and symptoms in an individual case Tinnitus headaches excessive warmth about the head and upper extremities in contrast to the colder feet vigorous pulsation in the neck head nodding with systole and even thyroid enlargement are all referable to the increased blood supply and hypertension in the upper parts of the body Conversely coldness of the legs numbness and pallor of the lower extremities and intermittent claudication are a reflection of the diminished vascular supply to the legs There is some considerable liability to subarachnoid haemorrhage Pain may be an important symptom It may be frankly exertional or it may be situated at sites where there are large collateral vessels Thus a case of Bramwell (1947) had pain in the right arm aggravated by using the limb and thought to be due to pressure on the brachial plexus by a dilated artery Evans (1933) was of the opinion that pain in the back could be produced by erosion of the ribs similar to the pain of vertebral erosion The fact that pain may occur in the absence of erosion of the ribs suggests that the origin of the pain may be due to pressure at the intervertebral foramen Similarly other rare symptoms may be attributable to a well developed collateral circulation Proganthism has been attributed to enlargement of the mandibular vessels (Mickerson 1947) A transverse myelitis at the level of the second dorsal vertebra was shown by

Haberer (1903) to be caused by a dilated anterior spinal artery Mental aberrations and transient palsies have been described There may be some relative over development of the tissues in the more vascular areas Intermittent claudication is not as frequent a symptom as might be thought In a personal case a woman of 45 complained of pain in her legs when she walked This pain had been present since childhood and was remarked upon at school Other examples are those of Parsons Smith (1921) and Woltman and Sheldon (1927) Enlargement of the thyroid with hyperthyroidism or without evidence of increased thyroid activity has been reported and some cases have been subjected to operation or other forms of treatment – Blackford (1928) Ulrich (1931) Amberg (1932) Brown (1934) Cookson (1936) Bramwell s case is particularly interesting because she presented rib notching with a constantly normal blood pressure Increased vascularity of the gland owing to the collateral circulation in which its arteries take part has been suggested as the cause of this increased thyroid activity It is not yet clear as to whether hyperthyroidism is a matter of chance or of increased vascularity and the experience of thyroid surgeons suggests that it is a very rare happening

The examination of a suspected case is most important and suspicion should be aroused in every case of hypertension in a young subject and in all cases of acyanotic congenital heart disease The most important clinical examinations are of the femoral pulse and a comparison of the blood pressure in the arms and legs in those cases where femoral pulsation is reduced or absent

The apparent absence of a femoral pulse even though there is a blood flow in the femoral arteries calls for comment Bonnet (1903) states that tactile appreciation of the pulse does not depend upon blood volume but on the sharp repercussions of the wave passing along the aorta Normally this is sudden and abrupt and is felt as a forcible pulsation When the blood reaches the aorta through numerous collateral channels this wave is damped the ascent of the wave becomes more gradual and the pulse correspondingly weaker Woodbury Murphy and Hamilton (1940) recorded pulse contours by a direct optical method from arteries above and below the coarctation Pulse contours above the constriction were normal in appearance below the constriction they were somewhat delayed flat and smooth This is in contrast to the normal state where as the pulse wave passes to the periphery the systolic pressure rises and the wave increase

Inspection reveals marked pulsation in the neck and perhaps head nodding with systole the latter particularly in the case complicated by aortic incompetence Enlarged tortuous pulsating vessels may be seen or felt in the scapular region and over the upper part of the body Pulsation may also be visible in the intercostal spaces and there may be visible pulsation of the internal mammary arteries Suzman (1947)

(fig 23) has shown that if the patient bends forward with his arms hanging down the *posterior intercostal spaces are opened up and dilated* intercostal vessels are rendered visible or more visible Further most of the phenomena of the collateral circulation can be emphasized by exertion or bending

Palpation shows the unmistakably arterial nature of the pulsation There may be a thrill over the dilated vessels or over the base of the heart and root of the neck Femoral pulsation is reduced or more often



FIG 23 Suzman s sign in coarctation of the aorta In the erect position few of the collateral vessels can be seen *On bending forward the number of visible arteries is greatly increased*

absent The upper extremities feel warmer than the lower and often look redder than the rather pale legs The heart may be normal in size or slightly hypertrophied Gross enlargement is present if there is aortic incompetence A systolic murmur may be heard over the heart loudest at the base It is heard over the back particularly well over the scapular vessels *and characteristically along both sides of the vertebral* column Not infrequently the murmur is inconspicuous over the heart but loud over the back and over the vessels of the collateral circulation A systolic murmur well to the right of the sternum over an intercostal space should evoke suspicion of coarctation Pulsation to the right of the sternum suggests a dilated aorta especially if accompanied by a ringing second sound There may be a diastolic murmur of aortic incompetence or the murmur of a patent ductus arteriosus Infective endocarditis may be accompanied by a diastolic murmur

The blood pressure in the arms is raised and is low in the legs a reversal of the normal physiological state Hypertension in the upper extremities is usually of the order of 150–200 mm systolic and at times even higher The pressure in the legs is markedly lower and may be unobtainable There is reason to suppose that this is quite often due to the difficult technical operation of obtaining a satisfactory reading in the leg whatever method is employed The direct intra arterial measurements of Steels (1941) showed the diastolic pressure to be elevated in the legs Normally there is synchronization of the radial and femoral pulses but in those cases of coarctation where the femoral pulse is present it is delayed Very occasionally the blood pressure in the arms is normal as in the case of Blackford (1928) King (1937) and in the two cases of Bramwell (1947) A normal blood pressure may be encountered in early childhood but a personal case had a systolic blood pressure of 160 in the arms at the age of two A child of Bramwell s with coarctation and subaortic stenosis had a blood pressure of 95/75 With these few exceptions the marked contrast in the status of the radial and femoral pulsation and the difference in blood pressure between the arms and legs is characteristic of coarctation and pathognomonic of aortic obstruction

Hypertension in Coarctation

Much has been written about the hypertension of coarctation and some hold that it is mainly mechanical in origin and due to the aortic stricture (Brotchner 1939) The results of operation are now cited in support of the view (Crafoord 1945) Page (1940) after experimental work in constricting the aorta of dogs concluded that there were grounds for considering the hypertension of coarctation to be of renal origin In fact the effects of aortic constriction are similar to the Goldblatt clamp Rytland (1938) held a similar view Steele (1941) who measured intra arterial pressure found the diastolic pressure to be raised in the legs in cases of coarctation and concluded that there was a general increase in arteriolar tone throughout the body similar to that in the common form of arterial hypertension The question of the cause of hypertension is of practical importance for if operation is designed to remove hypertension and its attendant dangers one must have certain knowledge of its mechanism In the light of modern knowledge it is unwise to dismiss the possibility of renal ischaemia as a cause and in this light it would be wise to perform operation at an early age before permanent and irreversible changes have set in

Coarctation at or above the Origin of the Left Subclavian Artery

This is a comparatively rare type of coarctation and only about twenty cases have been reported The constriction is situated at or just proximal to the origin of the left subclavian In some cases the left

subclavian artery may be congenitally narrowed at its orifice or in its length as part of a general abnormality of the aortic arch In rare cases East (1932) and Love and Holmes (1939) there may be an aberrant origin or an associated stenosis of the right subclavian artery When there is a constriction proximal to the origin of the left subclavian artery the left arm is excluded from the high pressure blood flow This has important clinical effects which may be recognized by asymmetrical development with overgrowth of bone and muscle of the right arm and right upper chest There is also disparity in the blood pressures of the two arms Thus in the case of Bayley and Holoubek (1940) the blood pressure in the right arm was 154/78 in the left arm 98/78 and in the left leg 90/80 The radial pulses were unequal in volume The collateral circulation may not be obvious but X ray examination may show rib notching which is confined to the right hemithorax If there is a widely patent ductus rib notching is absent

In the case of Love and Holms (1939) a negro of 44 the blood pressure was 210/95 in the left arm and 150/90 in the right arm *Autopsy showed that the left common carotid and innominate arteries* arose from a common trunk The right subclavian a very narrow artery arose from the innominate Coarctation was present distal to the origin of the left subclavian artery

RADIOLOGY The most important radiological sign is erosion of the under surfaces of the ribs first observed radiologically and correlated with anatomical observation by Railsback and Dock (1929) Meckel (1827) figured erosion on the ribs in a plate of his case of coarctation Jordan (1830) mentioned that in his case the ribs had become deeply absorbed and sulcated in some parts by the pressure of the branches of the (intercostal) arteries It seems strange that the best part of a century should elapse before there was radiological recognition of these anatomical changes When present the erosions are usually multiple and are found most often on the inferior surfaces of the *posterior portions of the third to the ninth ribs* (figs 21 and 24) Two types of notching are discernible either shallow or deep Both types are due to pressure atrophy of the rib where it comes in contact with the dilated tortuous intercostal vessel A loop of the vessel will cause a deep notch while a straighter vessel will cause a shallow depression (Wolke 1937) Although there may be marked tortuosity near the neck of the rib the vessel is here distant from the rib and notches do not *occur at this site* The process of pressure atrophy and erosion of bone is wholly comparable to that observed in pressure of an aneurysm upon bone elsewhere Most observers have considered rib notching to be pathognomonic of coarctation Laubry and Balsac (1937) have observed similar but less marked changes in hypertension and aortic incompetence Such notching does occur but it is trivial in degree Notching confined to the right ribs suggests an abnormal site of coarctation

Notching of the ribs is not invariably present in all cases of coarctation and it may not be pronounced It is absent in those cases where there is a widely patent ductus arteriosus (Evans 1933) In Bramwell s series it was present in seventeen cases but in nine it was so little marked as to be of no diagnostic value or completely absent Of these nine cases two were children and three cases had a patent ductus arteriosus In young subjects erosion of the ribs is comparatively rare the earliest recorded case being at the age of six



FIG 24 Coarctation of the aorta in female aged 30 Note the small aortic knuckle and the well marked notching of the inferior surfaces of the ribs

The heart may show slight enlargement with emphasis of the arc of the left ventricle Gross enlargement is observed when aortic incompetence or infection are complications The ascending aorta usually bulges to the right owing to pre stenotic hypertension although cases are described where it is small on account of hypoplasia These latter are rare and a vigorously pulsating dilated ascending aorta is generally observed In some cases dilatation of the aorta may be of aneurysmal proportion The aortic knuckle is small inconspicuous or absent but in some cases it has been large and prominent Often the shadow of the enlarged left subclavian artery is mistaken for an aortic knuckle The superior mediastinum is widened with vigorous pulsation on its left border which usually presents a convex appearance It may be difficult or impossible to visualize the aortic arch and the adjacent part of the descending aorta even in the oblique view This circumstance is a very suggestive finding (Fray 1930) The surgical experience of Crafoord explains the invisibility of these parts of the aorta as being due to the

ligamentum arteriosum retracting the aorta towards the mediastinum with the result that the aorta is surrounded by a less amount of air carrying tissues with corresponding diminution in its visibility with the barium filled oesophagus Wolke (1937) has shown that in the antero posterior position there is an indentation of the oesophagus corre sponding to the portions of the aorta above and below the constriction Rotation of the subject 25 or 30 to the right may render this more clearly visible

Visualization of the descending aorta in the oblique position may be extremely difficult if not impossible It may be possible to demon strate a gap or narrowing of the arch This is an unreliable sign for a number of cases fail to show any marked external constriction of the aorta despite the presence of a diaphragm within its lumen If however a gap is observed it will be seen that pulsation below the gap is diminished or absent and in marked contrast to that above the con striction Laubry and Balsac (1937) and others using radiokymo graphic methods have been able to show the site of constriction of the aorta This method if successful graphically illustrates the diminished pulsation of the aorta distal to the constriction Tomography suggests itself as a likely method to employ but it has rarely been successful (Bramwell) Bramwell has noted in cases where rib notching is absent as in those where the ductus remains patent a double aortic knuckle the upper arc being formed by the left subclavian artery and the lower by the blind end of the descending aorta

Angiocardiographic examination has been successfully employed by Grishman Steinberg and Sussman (1941) and Chavez *et al* (1947) to demonstrate the site of coarctation

ELECTROCARDIOGRAM There are no characteristic changes in the electrocardiogram (fig 25) which is often physiological Ultimately as might be expected there is the picture of left ventricular hypertrophy A normal axis deviation does not preclude a diagnosis of coarctation Auricular fibrillation is sometimes present

COURSE AND PROGNOSIS Many patients are robust and active until middle life when the symptoms discussed above appear and are progressive The difficult time is the third decade when men work and play hardest and women bear children In general it may be stated that if the stenosis is severe and symptoms appear early or if they date from childhood the prognosis is poor Patients who are symptom free at the age of 30 have a more favourable outlook A large number of cases die in infancy and these are usually of the infantile type of coarctation and associated with other grave anomalies of the heart

It is very difficult to assess the prospects of the individual case Some have held that coarctation with a fully developed collateral circulation places no demands upon the cardiac reserve and that cardiac hyper trophy was absent in the uncomplicated case This may be true of some

cases but the post mortem experience of Abbott (1928) who found cardiac hypertrophy in three quarters of her cases suggests that there is a definite strain imposed by the lesion This strain although con stantly present may not be sufficient to cause trouble in the first half of life as long as the coarctation is uncomplicated Severe physical strain or intercurrent infection may however precipitate failure Blackford (1928) mentions sudden death from rupture of the aorta during strenuous exertion The average age at death in Abbott s series

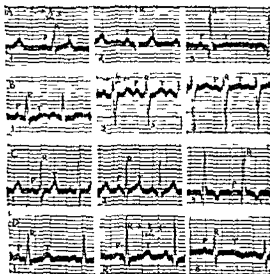


FIG 25 The electrocardiogram in coarctation of the aorta The four cases illustrated are below the age of 16

was 32 with extremes of 3 and 92 To judge from reported cases about 25 per cent of all cases die before the age of 20 half of the cases do not live longer than 40 years and nine tenths of the cases die before the age of 50 Thus only a tenth of the cases pass the age of 50 The causes of death in order of frequency as given by Maud Abbott are congestive heart failure rupture of the aorta or heart cerebral haemor rhage and bacterial endocarditis Reifenstein Levine and Gross (1947) have analysed 104 cases collected since Abbott s (1928) review and found the average age at death to be 30 5 years Sixty one per cent of these cases died during or before their fortieth year The majority of deaths from rupture of the aorta or from cerebral haemorrhage occurred in the second and third decades from bacterial endocarditis or aortitis in the first five decades and from congestive failure in the

third to fifth decade In order of frequency 26 per cent died of incidental causes unrelated to their anomaly 23 per cent from rupture of the aorta 22 per cent from bacterial endocarditis 18 per cent congestive heart failure and 11 per cent from intracranial lesions

DIAGNOSIS The condition is probably much more common than is suspected and careful examination including the femoral pulse should be made of every young subject with hypertension and in particular those referred as cases of congenital heart disease or aortic incompetence The presence of thrills and murmurs in atypical situations such as those found over the collateral vessels the occurrence of intermittent claudication in the young and marked disparity between the temperatures of the extremities are all circumstances worthy of critical consideration

In Abbott s (1928) collected series of 200 cases an ante mortem diagnosis had been made in 14 per cent Reifenstein s (1947) series showed an ante mortem diagnosis of 40 per cent so there is reason to suppose that the clinician is becoming aware of the abnormality

The cardinal features which should be constantly borne in mind in making a diagnosis are an absent or diminished femoral pulse hypertension in the arms and a lower blood pressure in the legs a collateral circulation with rib erosion in the X ray picture systolic bruits heard in atypical situations along the spine or over the vessels of the collateral circulation

TREATMENT Medical treatment is of a prophylactic nature and is concerned with the avoidance of strain and protection from infection according to the individual needs of the patient Focal sepsis in teeth or tonsils should be dealt with and it is wise to give penicillin befor and after any operation to the teeth or tonsils

Surgical treatment of coarctation was first considered by Blalocl (1944) There are two possible surgical procedures Firstly the lef subclavian artery can be used to by pass the stricture its divided enc being anastomosed to the aorta distal to the stricture Such an operatior might have a possible application in some cases of the infantile type oi coarctation It might well be the operation of choice where the aorta is diseased or where there is difficulty in freeing the aorta sufficiently to excise the stricture and suture the divided ends without undue tension Such operations have been performed by Blalock (1947) and Clagett (1947) Secondly the stricture may be excised after mobilization of the aorta and the divided ends joined together This operation was successfully performed in 1944 by Crafoord and Nylin and by Gross (1945)

The avowed reason for operation is the poor individual prognosis of coarctation with its rather high death rate up to the age of 40 If the operation is to be performed for the relief of hypertension and its attendant dangers it would seem desirable to perform it as early as

possible ideally before the age of 20 This minimizes the possibility of the permanent effects of renal ischaemia having set in Apart from this degenerative changes are common in the aorta and these may increase the technical hazards of operation in older subjects Even if operation is performed the point should not be lost sight of that a bicuspid aortic valve is present in a large number of cases an anomaly only second to the patent ductus arteriosus in its risks of infective endocarditis Equally there remains the berry aneurysm which may rupture even in the absence of hypertension

The results of operation are impressive Crafoord has performed the operation on twenty cases with two deaths one from haemorrhage from an intercostal artery and the other from a dissecting aneurysm The immediate effects of operation are a sudden fall of the blood pressure level in the arms and a rise in pressure in the legs Pulsation becomes evident in the arteries of the legs and the legs become warm Equalization of pressure in arms and legs comes after a period of time often extending over a period of months The collateral vessels disappear gradually

It is early to pass final judgment on this operation but as far as our present knowledge goes it is a major advance in physiological surgery and must in many young cases be confidently recommended to the sufferer with coarctation

Congenital Stenosis of the Aorta Distal to the Isthmus

(Syn Congenital Stenosis of the Abdominal Aorta)

Whilst coarctation of the aorta is comparatively common congenital stenosis of the aorta distal to the isthmus is extremely rare A con genital constriction may occur between the first and second intercostal arteries (Costa 1930) just above the diaphragm (Hasler 1911 Schleckat 1933) proximal to the renal arteries (Steele 1941) and distal to the renal arteries (Power 1861 Maycock 1937)

The origin of such a constriction is obscure Normally fusion of the dorsal aortae commences at about the fourth week and ultimately a single channel the thoracic and abdominal aorta is formed Incom plete fusion may result in a central thin partition to the descending aorta It has been suggested by Maycock (1937) that in stenosis distal to the isthmus there is a failure of the normal fusion of the dorsal aortae and subsequent disappearance of one of them

The degree of stenosis may be only slight or may be complete Severe stenosis or complete obliteration of the aorta gives rise to aneurysmal dilatation in the aorta proximal to the seat of obstruction and there is also a tendency to thrombosis at the site of the lesion It is possible for lesions other than congenital to produce a stenosis or even obliteration of the aorta The proof of a congenital origin of the stenosis lies in the presence of a collateral circulation developed by

compensatory hypertrophy of the intercostal and internal mammary vessels in conjunction with the superior and inferior epigastric vessels For a complete discussion of these cases the reader is referred to the work of Maycock

There is no typical symptomatology of the condition and such symptoms as are present in the few recorded cases are similar to those of coarctation of the aorta The lesion may be entirely latent There may be attacks of dyspnoea and palpitation and a sense of fullness and pressure in the chest Epileptiform convulsions were present in two cases

The physical signs are those of cardiac enlargement hypertension and the presence of a collateral arterial circulation

Death may result from intercurrent infection cardiac failure or rupture of a blood vessel The average age at death in six recorded cases was 32 with extremes of 15 and 49

Interruption of the Aortic Arch in its Distal Portion with Hypoplasia of the Aorta This is a rare abnormality and Abbott (1927) analysed five cases Evans (1933) reported three cases in his series A recent well documented case is that of Stewart (1948)

The principal anatomical feature is the abrupt termination of the aorta at the level of the left subclavian the arch of the aorta being continuous with this latter vessel The pulmonary artery after giving off a right and left branch continues by a widely patent ductus arteriosus into the descending aorta There may be associated abnormalities of origin of the left subclavian which may be given off as the first branch of the descending aorta and in one of Evans s cases both subclavians arose from this site In Stewart s cases there was a small interventricular septal defect The interruption of the arch is so complete that there is no fibrous band or any communication between the aortic arch and the de cending aorta (fig 26)

The embryological origin of the anomaly is by the abnormal disappearance of the distal part of the fourth left aortic arch and of the left dorsal aorta between the fourth and last left aortic arch The fourth left arch continues as the left subclavian artery There is thus interruption in the arch of the aorta the descending portion being absent and the ductus becomes continuous with the descending aorta

This anatomical arrangement were the subject to survive sufficiently long would be reflected by cyanosis in the lower half of the body and a good colour in the upper half Death is usual in earliest infancy but Abbott (1927) mentions survival of a case to five and a half years

Congenital Absence of the Ascending Aorta These cases are rare and Abbott (1927) has analysed ten To these may be added the case of Evans (1933) which in addition was an example of a cor biloculare The ductus arteriosus is widely patent and continues undiminished in calibre into the descending aorta Slightly proximal to

the point of junction of the ductus and aorta a common trunk is given off from which arise the vessels of the arch (fig 26)

Abnormalities of the Right Subclavian Artery

The right subclavian artery may arise distal to the left subclavian as the last branch of the aortic arch To reach the right side it passes behind the oesophagus but it may also pass between the trachea and oesophagus or anterior to the trachea Rarely it may arise from the descending aorta below the ligamentum arteriosum

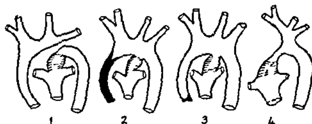


Fig 26 Types of stenosis and atresia of the aortic arch (After Evans) 1 Interruption of the aortic arch in its distal portion 2 Atresia of the ascending aorta ductus widely patent 3 Atresia of the aortic orifice 4 Absence of the ascending aorta

An abnormal right subclavian may give rise to symptoms Bayford (1794) reported dysphagia in a case with an anomalous right subclavian artery passing between the trachea and oesophagus and Holzapfel (1899) refers to the even earlier cases of Murray (1768) and Brewer (1791) In Murray s case the artery was retro oesophageal Most often the condition remains symptomless and unrecognized The abnormal artery may be injured accidentally as in the case of Kirby (1818) where it was injured by a piece of bone penetrating the oeso phagus a probable factor in the subsequent death of the patient

Hypoplasia of the Aorta

Maud Abbott (1927) defines hypoplasia of the aorta as that condition in which the lumen of the arterial vessels in the greater circulation remains abnormally small and the walls unnaturally thin and elastic It appears to have been first recognized by Morgagni (1788) Rokitansky (1844) noted the occurrence of an infantile aorta with associated cardiac hypertrophy and genital hypoplasia Virchow (1872) noted a narrow aorta and cardiac hypertrophy Spitzer (1897) com mented upon the same association and stated that only if the subject lived to adult age would cardiac hypertrophy ensue There are more than a hundred cases in the literature and discussion has centred round whether it is a true congenital abnormality or whether it is due to

factors operating after birth It is so *frequently associated with* lymphoid and thymic hypoplasia that it may be the expression of a constitutional type (Philpott 1929)

Hypoplasia of the aorta may occur as an isolated condition or as a complication of other congenital cardiac defects This suggests that cases might well be divided into these two groups As an isolated defect it is purely developmental and is often associated with other visceral abnormalities notably of the genitalia or with thymic disturbance The case of Cluver and Jokl (1942) presents points of interest An *athlete aged 32 collapsed* and died after a game of Rugby football Post mortem examination showed a hypertrophied heart with a normal sized but very *thin walled ascending aorta and aortic arch* The descending aorta was less than half the normal size Atheroma involved the orifice of the left coronary artery and some patches were present in both right and left coronary arteries There was in addition a left hydronephrosis and a persistent thymus The spleen was enlarged and all lymphoid tissues were hyperplastic His identical twin brother had died during exertion at the age of 30 It was held that persistence of the thymus had arrested the normal puberty and post pubertal development of the circulatory system so that the descending aorta remained in an infantile state This increased resistance led to cardiac hyper trophy and dilatation and finally resulted in a fatal circulatory crisis Valentine and Nicholl (1945) *report the case of a man of 31 who pre* sented a hypertrophied heart with hypoplasia of the aorta and extensive genitourinary abnormalities No mention is made of the lymphoid tissues Other examples are those of Whittle (1929) Ikeda (1932) and Werley Waite and Kelsey (1944) Cooke and Cloake (1943) report two cases Their first case a diabetic male of 31 had extreme cardiac enlargement with moderate hypoplasia of the descending and abdominal aorta and pituitary hyperfunction was suspected In their second case a 35 year old female with acromegaly there was gross cardiac enlarge ment *predominant hypertrophy of the left ventricle an interauricular* septal defect and hypoplasia of the aorta

The foregoing brief account indicates that there is a quite definite type of aortic hypoplasia with either lymphoid or endocrine changes and often with abnormalities of the genitourinary tract The hypoplasia need not necessarily involve the whole length of the aorta but even in those cases where hypoplasia is most apparent distal to the aortic arch comment has been made on the soft thin walls *of the ascending* aorta

Hypoplasia of the aorta as *a secondary abnormality is the almost* constant concomitant of significant defects of the auricular septum It may *complicate* the va ous types of stenosis in the aortic tract or be present when mitral stenosis is established at an early age Balfour (1898) insisted upon the preponderant role of early mitral stenosis in

the genesis of aortic hypoplasia He supported the view of a congenital mitral stenosis and his ideas were sufficiently advanced for him to state that rheumatic involvement of the mitral valve always entailed some degree of stenosis For him hypoplasia of the aorta was a mechanical effect of mitral stenosis because less blood was transmitted to the aorta As a consequence the normal distension of the aorta was lessened and growth and metabolism of tissues was impaired Autopsy experience has not substantially confirmed that there is marked hypo plasia of the aorta in rheumatic mitral stenosis In coarctation of the aorta the element of hypoplasia may be overlooked as it is often obscured by secondary dilatation of the ascending aorta

The anatomical specimen in all types of aortic hypoplasia shows reduction in the calibre of the aorta as a whole or of the segment distal to the arch The walls are thin and abnormally soft and elastic and their changes may extend to the branches The intima is smooth and most often healthy Microscopically there is a diminished breadth of all three layers but especially of the media

The effects of hypoplasia have been much discussed particularly in its relationship to enlargement of the heart As many of the cases have been victims of sudden death there is a paucity of records in regard to the blood pressure In many of the well studied cases the blood pressure has been normal but hypertension cannot always be ruled out The experimental work of Eyster (1927) who temporarily constricted the aorta of dogs showed that hypertrophy of the heart followed some months later Cooke and Cloake suggest that a possible dilatation of the heart to cope with extra output requirements in strenuous work might act as the stimulus for hypertrophy Even if this were so it is difficult to explain the gross enlargement of the heart that is present Some (Amsler 1912 Courville and Mason 1938) suggest an endocrine factor and arguing from the incidence of cardiac hypertrophy and failure in the absence of hypertension in acromegalics find reason to suppose that a glandular element enters into the matter Clearly here is a matter that requires patient investigation for the actual cause of hypertrophy be it increased work or glandular activity remains hidden

derived from the distal part of the fourth left root arch may arise from the aortic root and pass behind the trachea and oesophagus In other cases it may arise from a persistent proximal portion of the fourth left arch and pass upwards to its distribution in front of the trachea and oesophagus

In the unique case of Beavan and Fatti (1947) a female child of 9 the aortic arch ascended into the neck to the level of the cricoid cartilage (fig 28) It then descended and passed backwards and to the left



Fig 29 High aortic arch Male aged 2[illegible] with no symptoms *Abnormality picked up at mass radiography and a mediastinal tumour was suspected until operation (Mr N R Barrett s case)*

behind the oesophagus and descended in a normal way A left subclavian artery arose from the descending aorta and a short patent ductus arteriosus entered the left subclavian thus completely encircling the trachea and oesophagus The child complained of dysphagia and presented a pulsatile swelling in the right side of the neck This was taken to be an aneurysm and ligatured with fatal results This anomaly results from formation of the aortic arch from the third embryonic arch instead of the fourth Another example of a high aortic arch is depicted in fig 29 and was in this case quite symptomless

RIGHT SIDED AORTIC ARCH Under this heading are included right sided aortic arch right sided aortic arch with persistent left root (Arkin) and double aortic arch

ANATOMY The aorta arches over the right bronchus passing to the right side of the trachea and oesophagus Two alternative courses may then be followed Either it descends on the right side crossing over at a lower level to reach the aortic opening in the diaphragm or in other cases immediately after crossing the right bronchus it passes behind the trachea and oesophagus widening into a diverticulum (persistent left root) which pushes these structures forward It may then descend on the left side to the diaphragm or distal to the diverticulum it may curve back to the right side and descend laterally to the ascending aorta gradually crossing to the left at about the level of the eighth or ninth thoracic vertebra

It is thus evident that a right aortic arch with a descending aorta on the right side is likely to be symptomless because its course is lateral to the trachea and oesophagus It is however possible that the arch of the aorta could exert pressure on the right upper lobe bronchus or the left innominate or left common carotid artery might be stretched across the trachea and so give rise to symptoms A right aortic arch with a left descending aorta is much more likely to cause symptoms because of its retro oesophageal course and because the ductus or ligamentum arteriosus completes a ring round both the trachea and oesophagus Even in the presence of a vascular ring of this type symptoms may not be present until such time as degenerative changes appear in the aorta

In the right aortic arch with a right descending aorta the first vessel given off the arch is a left innominate artery followed by a right common carotid and a right subclavian artery

CLASSIFICATION AND TYPES An embryological classification would include most if not all, of the types in the category of a double aortic arch The anatomical types described are *mainly differentiated by the* degree of persistence of the left fourth arch from its most attenuated *form down to its* most obvious form in double aortic arch Cases with a persistent left root occupy an intermediate position in this scale *and Arkin (1936)* consider that the fourth left arch undergoes stenosis in the isthmus region The combination of left subclavian artery short occluded vessel *and persistent left root* comprising the fourth left arch forms a tight ring round the trachea and oesophagus and leads to the persistence *of the fourth* right arch (figs 30 and 31)

The development of a right sided aorta is more easily understood when it is *considered* in relation to other abnormalities of the fourth arch *A glance at the table adapted from* Roche Steinberg and Robb (1941) makes this reasonably clear

The types of right sided aortic arch are as follows

(1) In addition to persistence of the right fourth arch there is persistence *of the* proximal portion of the left fourth arch The aorta crosses the right bronchus and the left subclavian (left arch) arises from

the front of the aorta and runs in front of the trachea to reach the left side The ductus arteriosus arises either from its usual site or from the left subclavian It may encircle the trachea and oesophagus and pull the right aortic arch behind these structures

(2) The right aortic arch has a similar origin to the preceding There is persistence of the distal portion of the left arch resulting in a diver

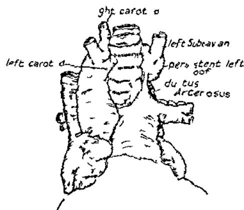


Fig 30 Anatomical picture in right aortic arch with persistent left root (Specimen R C S Museum)



Fig 31 Right aortic arch (After Arkin)

ticulum at the end of the arch the persistent left root of Arkin The left subclavian arises from the diverticulum and runs behind the trachea and oesophagus The ductus arteriosus joins the diverticulum The position of the diverticulum behind the trachea and oesophagus leads to these structures being pushed forward a point of radiological importance

(3) A true double aortic arch arises from persistence of both the

right and left fourth arches It is often associated with congenital cardiac anomalies that are incompatible with life It may occur as a sole abnormality The aorta ascends to the right and divides into two limbs (fig 32 A and B) A posterior limb passes behind the oesophagus and the anterior limb runs to the left in front of the trachea These two vessels which may be large or unequal in size merge together again in the left side of the mediastinum beyond the insertion of the ductus to form the descending aorta The vessels of the arch may come from either limb or from both limbs Commonly the posterior limb gives rise to the right subclavian and the right common carotid arteries and the anterior limb to the left subclavian and left common carotid artery The trachea and the oesophagus are thus encircled and from the surgical point of view there may be occasions when it may be impossible to divide the ring without interfering with important vascular channels (Gross 1945) The encirclement of the trachea and oesophagus may influence the clinical picture at an age when degenerative changes are apt to take place in the vascular components of the ring

ANOMALIES OF THE AORTIC ARCHES CONSIDERED IN RELATION TO A FUNCTIONING DOUBLE ARCH

An ingenious classification and explanation has been propounded by Edwards (1948) As a reference pattern he takes a functioning double aortic arch In the young embryo the ventral aorta gives off two arches which encircle the primitive alimentary canal and then join to form the dorsal aorta By atresia or disappearance of certain parts of this system the normal aortic pattern is produced as well as anomalies of the arches In group 1 the ductus and descending aorta are approximately in their normal positions Group 2 anomalies are the mirror image of those of group 1

GROUP 1 LEFT SIDED DUCTUS ARTERIOSUS AND DESCENDING AORTA

(1) *Functioning Double Aortic Arch*

The types illustrated differ only from each other in the diameter of the anterior (left arch) a narrow anterior arch being the more common The anterior (left) arch passes in front of the trachea The posterior (right) arch passes over the right bronchus and behind the oesophagus the two arches meeting either behind the oesophagus or to the left of the midline The trachea and oesophagus are thus completely encircled by a vascular ring rendered taut by the ligamentum arteriosus (fig 32 A and B)

(2) *Partial Atresia of Double Aortic Arch*

Atresia has occurred between the origin of the left subclavian and the aortic insertion of the ductus arteriosus This is a not uncommon type and has been described by Arkin (1936) (fig 32 C)

(3) *Right sided Aortic Arch with Retro-oesophageal Segment and Left*

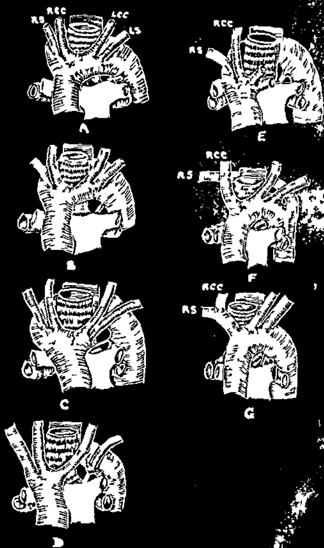


FIG 32 Abnormalities of the aortic arch A and B double aortic arch C partial atresia of double aortic arch D right sided aortic arch with persistent left root giving origin to left subclavian artery E right sided aortic arch with left subclavian arising from an innominate artery F left aortic arch with retro-oesophageal right subclavian artery G normal type of arch (After Edwards)

sided Descending Aorta Left Subclavian Artery originating from an Aortic Diverticulum

Atresia has occurred in the region between the left common carotid and left subclavian arteries The left subclavian arises from a diverticulum (representing the posterior portion of a left aortic arch) ductuc arteriosus attached to the diverticulum (fig 32 D)

(4) *Right sided Aortic Arch with Retro oesophageal Segment and Left sided Descending Aorta Left Subclavian from a Left Innominate Artery*

A gap exists between the origin of the left subclavian artery and the aortic insertion of the ductus The ductus is commonly inserted into a diverticulum formed by a remnant of the left arch (Arkin 1936) (fig 32 E)

(5) *Left sided Aortic Arch and Left sided Descending Aorta Right Subclavian Artery arising from the Distal Arch*

The right portion of a double arch has disappeared between the origins of the right common carotid and the right subclavian arteries The right subclavian arises as the fourth branch of an otherwise normal aorta and passes behind the oesophagus (fig 32 F)

(6) *Left sided Aortic Arch and Left sided Descending Aorta Normal pattern*

That part of the posterior arch distal to the origin of the right subclavian artery has disappeared to produce a normal arrange ment of the vessels (fig 32 G)

(7) *Hypothetical form yet to be described*

Right sided aortic arch and left sided descending aorta with atresia of the left aortic arch between the origins of the left common carotid and left subclavian arteries

GROUP 2

(1) *Right sided Ductus Arteriosus with upper part of Descending Aorta on the right side Right Subclavian from an Aortic Diverti culum*

A rare type A case has been reported by Edwards (1948) (fig 33 A)

(2) *Left sided Aortic Arch with Retro oesophageal Segment and upper part of Descending Aorta on the right side Right Subclavian from the Right Innominate Artery*

The part of the right arch between the origin of the right sub clavian and the aortic insertion of the ductus has disappeared (Paul 1948) (fig 33 B)

(3) *Right sided Aortic Arch with upper part of the Descending Aorta on right side Left Subclavian from the Aortic Arch of Descending Aorta*

The left arch between the origin of the left common carotid and the left subclavian disappears (Lockwood 1884) (fig 33 C)

(4) *Right sided Aortic Arch with upper part of the Descending Aorta on the right side Branches of the Arch a mirror image of normal* A frequent arrangement in the tetralogy of Fallot with right aortic arch (fig 33 D)

(5) *Hypothetical forms*

(*a*) Functioning double aortic arch with ductus on the right side

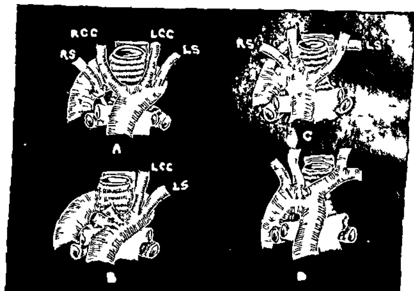


FIG 33 Abnormalities of the aortic arch A left sided aortic arch with descending aorta *on the right side Right subclavian* arises from an aortic diverticulum B similar type to A with right subclavian arising from an innominate artery C right sided aortic arch with retro-oesophageal left sub clavian artery D *right aortic arch in the tetralogy of Fallot (After Edwards)*

(*b*) Partial atresia of right arch between the origins of the right common carotid and right subclavian

(*c*) Atresia between origins of the right subclavian artery and the aortic insertion of the ductus

It will be noted that these anomalies are mirror images of those already known and belonging to group 1

INCIDENCE *The right sided aorta has hitherto been regarded as a* rare congenital malformation In Abbott s (1931) one thousand cases it occurs only *fourteen times* The anomaly was first described by Sandifort (1793) The association of a right sided aorta with the *tetralogy of Fallot* has been known since the time of Corvisart (1818)

Assmann (1924) who first described the radiological picture although incompletely referred to it as Hohe rechtslage der Aorta and from its occurrence in his three cases with other congenital anomalies he assumed that it was always combined with these latter A large atrial septal defect was present in the case of Assmann Bedford and Parkinson (1936) reported its association with the Eisenmenger Complex and it occurs in 50 per cent of cases of persistent truncus arteriosus It is found with the various types of dextrocardia It is probably much more

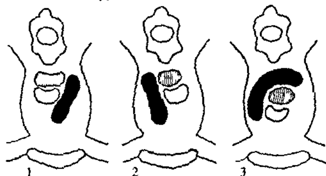


Fig 34 Diagram of the types of right sided aortic arch (After Brumlik) 1 Normal 2 Assmann s type 3 Arkin s type with persistent left root

frequent than is at present estimated and no doubt the mass radiological surveys of the population will bring many symptomless cases to light

In regard to the double aortic arch most of the twenty cases collected by Lockhart (1929) were found in children under seven months of age In Lian and Marchal s (1937) case there was a functioning double aorta at the age of 33

CLINICAL PICTURE Most often the anomaly is symptomless and only found at post mortem or in the dissecting room A large ring is unlikely to give rise to symptoms but a small tight ring may cause tracheal compression and less frequently oesophageal obstruction with dysphagia

Dysphagia may occur in adults after 50 when degenerative vascular changes take place When present dysphagia is due to pressure on the oesophagus by the aorta and its left root (fig 34) or by the posterior limb of a double aorta Occasionally when an anomalous left subclavian pursues a retro oesophageal course it may also cause dysphagia This general type of dysphagia has been called dysphagia lusoria According to Fagge (1886) the term lusoria (from the Latin *lusus* a freak) was first applied by Bayford of Lewes (1789) to a case of dysphagia where an anomalous right subclavian artery passed between the trachea and oesophagus Arkin (1936) has called attention to certain

physical signs present in his cases These are dullness along the upper right border of the sternum extending *as high as the right sterno* clavicular articulation visible and palpable systolic pulsation in the second *and third right* spaces near the sternum and in the right supra clavicular fossa maximum intensity of the aortic sounds to the right of and above their usual situation The other heart signs are normal The diagnosis is essentially radiological and in the absence of symptoms may be purely accidental



FIG 35 Tracings *of the right sided aortic arch (After Bedford and* Parkinson) A with persistent left root B Assmann s type

In regard to the special symptomatology of the double aortic arch there may be no symptoms in adults *In children the symptoms are* those of noisy breathing stridor cyanosis and dysphagia with exag geration *of these symptoms* when the child is fed or when it lies down Recurrent tracheitis and bronchial infection is common and the case may terminate in broncho-pneumonia as did that of Snelling and Erb (1933) *From a diagnostic point of view wheezing from birth and* later dysphagia are the most suggestive symptoms Actual diagnosis is mainly *radiological and so far the bulk of cases have only been recog* nized at autopsy Awareness of the possibility of a double aortic arch and its importance from the point of view of surgical correction should lead to more frequent diagnosis in the future

RADIOLOGY The aortic knob is absent on the left side and the aorta projects to the right where it may be seen either as a definite knob below the right sterno clavicular joint or as a band like shadow extending up to the *right clavicle The appearance with barium* in the oesophagus is pathognomonic as the right sided aortic arch lies to the right of the oesophagus which latter is indented *and pushed to the* left There may sometimes appear to be two aortic knobs the smaller and less clear one being on the left side (fig 35) This left sided knob is the persistent left aortic root and being placed behind the oesophagus is not so clearly visible The trachea is also placed to the left with the

result that the left bronchus tends to become vertical as the continuation of the trachea whilst the right bronchus arises at an obtuse angle This reversal of the normal condition has been described by Lian and Marchal (1937) as the sign of inversion of the bronchi These same observers have noted a functional double aorta in which both right and left aortic knobs were actively pulsating Where the right sided arch is combined with the tetralogy the aorta appears larger than normal and displaced to the right

In the right oblique position the right aortic arch and diverticulum push the trachea and oesophagus forward giving a very characteristic picture Absence of displacement of the oesophagus in this view suggests that there is no aortic diverticulum Examination of the patient with lipiodol in the trachea and a barium swallow may show narrowing of the trachea above the bifurcation and some slight indentation of the oesophagus (Gross 1945)

A practical point if surgery is entertained is that the ductus arteriosus is usually situated on the same side as the upper portion of the descending aorta and so determines the side on which the chest should be explored

Angiocardiography is of value in tracing the aorta and its use is well exemplified in the case of Roche Steinberg and Robb (1941) In this case a large pulsating shadow on the right border of the heart was thought to be an aneurysm but this method showed it to be a tortuous descending aorta the continuing part of a right sided aorta with a presistent left root

COURSE AND PROGNOSIS The right aortic arch unless combined with other anomalies may pursue an entirely symptomless course The chief risks are theoretical and would be engendered by oesophagoscopy or other intrathoracic operations The anomaly of itself may not shorten life and the prognosis is that of the associated congenital heart abnormality if any

Surgical Treatment of Double Aortic Arch

The first successful operation was performed by R E Gross on 9th June 1945 The patient was an infant of one year with tracheal obstruction and recurrent tracheitis Preliminary investigation had disclosed compression of the trachea above the bifurcation and indentation of the posterior wall of the oesophagus

Under cyclopropane anaesthesia the chest was opened through a left anterolateral incision through the third interspace the second and third costal cartilages being divided When the mediasternal pleura was opened dissection of the tissues offered a clear view of the great vessels all of which were identified In this case there was a double aorta the aorta dividing just above its origin into two limbs which passing respectively anterior and posterior to the trachea joining to

form the descending aorta thus making a ring round the trachea and oesophagus The ring was tightened by the ligamentum arteriosum which ran from the left pulmonary artery to the point of junction of the two aberrant vascular limbs of the double aorta

The tracheal compression was relieved by division between silk ligatures of the ligamentum arteriosum This caused the pulmonary artery to fall forward away from the trachea This procedure did not cause complete relief and so it was decided to interrupt the smaller anterior limb in such a manner that the circulation to the descending aorta would not be compromised The anterior limb was accordingly severed between ligatures between the left common carotid artery and the left subclavian The left common carotid immediately displaced itself superiorly and to the right and the trachea was obviously relieved of pressure The chest wall was then closed the left lung being com pletely expanded before final suture of the wound

Post operative treatment consisted of a steam room for one week and parenteral sulphadiazine and penicillin for sixteen days At the end of a week there was no pyrexia and no distress of any kind

It appears from a surgical point of view that the essential procedures are division of the smaller arch and of the ligamentum arteriosus In those cases of right aortic arch where there is actual compression division of the ductus may enable the pulmonary trunk to fall away from the trachea An abnormal subclavian artery should be ligatured and divided and the ligamentum arteriosus may be divided as well

Abnormalities of the Coronary Arteries

Abnormalities in the origin of the coronary arteries are uncommon They have been studied by Bland White and Garland (1933) and informative reviews are given by Soloff (1942) and Kaunitz (1947) The left coronary artery is most frequently involved perhaps because the ostium of the left coronary artery is much closer to the pulmonary sinus than is that of the right Soloff mentions seventeen cases of anomalous left coronary artery and to these may be added that of Proescher and Baumann (1944) and the two cases of Lyon Johansmann and Dodd (1946) The recent survey of Kaunitz lists twenty seven cases

The effect of such an anomaly where placement of a coronary artery in the pulmonary artery results in a qualitative defect in the blood supply is to cause degenerative changes with replacement fibrosis in the territory of the artery derived from the pulmonary artery Dilatation of the affected chamber ensues and the heart hypertrophies The lesion has been recorded as a cause of the so-called idiopathic hypertrophy of the heart (page 42) In some cases if survival has been long enough an arteriovenous aneurysm may develop in the heart wall (Abbott 1927 Brooks 1885) In one case that of Biorck and Crafoord (1947) an anomalous branch of the left coronary artery entered the pulmonary

artery forming an arteriovenous aneurysm with a continuous bruit Occasionally the right coronary or a third coronary artery the latter of little significance may arise from the pulmonary artery There may be a single coronary artery In complete transposition of the vessels both coronaries arise from the transposed aorta and venous ventricle

PATHOLOGY When a coronary artery arises from the pulmonary artery it results in a qualitative defect in the blood supply leading to degenerative changes and replacement fibrosis in the territory supplied by the aberrant vessel Only when an accessory coronary arises from the pulmonary artery are these changes minimal and negligible In the two cases where the right coronary artery arose from the pulmonary artery Monckeberg s (1914) man of 30 who died of a dural haemor rhage and Schley s (1925) man of 61 who had syphilis there were no gross changes in the musculature of the right ventricle This is in marked contrast to the case where the left coronary arises from the pulmonary artery It seems possible therefore that the right ventricle which performs less work than the left may be able to function with a less oxygen supply than the left ventricle It is interesting to note that in the ordinary human subject arteriosclerosis occurs with about equal frequency in both coronary arteries yet it is uncommon to find marked degenerative changes and fibrosis in the right ventricle This observation suggests that the ogygen needs of the left ventricular musculature exceed those of the right ventricle which latter can function in the normal case with the low oxygen content of venous blood In this connection too it should be remembered that the right ventricle is richly supplied with Thebesian vessels and infarction of the lateral wall of the right ventricle is uncommon in comparison with infarction of the left ventricle due to the collateral circulation provided by these vessels King (1941) suggests that changes in the left ventricular wall are not necessarily due to the low oxygen content of the venous blood that it receives He believes that anastomosis between the coronary vessels are such that the affected area receives its blood supply from the unaffected coronary artery and that flow in the aberrant vessel is toward the pulmonary artery rather than from it When degenerative changes occur they are due to failure of the collateral circulation under stress The frequent onset of symptoms when the infant passes from the vegetative to the more active state lends some support to this view

The actual degenerative changes are peculiarly constant in the recorded cases All stages of degenerative disease are present including fatty changes ischaemic necrosis fibrosis and calcification The wall of the aberrant coronary artery is often thin and vein like Soloff (1942) has paid particular attention to the embryonic sinusoids which main tain a communication between the left coronary and the ventricular

cavity These were numerous in his case an infant of four months and appeared as large distended spaces in the left ventricular musculature which were lined with a single layer of flat endothelial cells Before the appearance of the coronary arteries these spaces carry blood to the muscle fibres and their persistence in numbers has been also commented upon by Grant (1926) and Bellet and Gouley (1932) The normal fate of these sinusoids is that as the coronary arteries are developed they are reduced to capillaries and persist as part of the

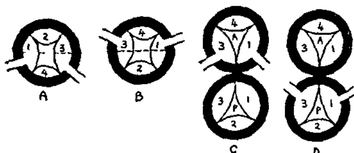


FIG 36 Changes in the distal bulbar swellings and coronary orifices A before rotation B after rotation C after normal division D after abnormal division so that coronary arteries arise from pulmonary artery

coronary circulation Some are destined to become the veins of Thebesius communicating with the ventricular cavity (Grant 1926) An aneurysm of the left ventricle was present in Barnard s (1938) case

EMBRYOLOGY The rudiment of the coronary artery or coronary bud appears about the fourteenth day of foetal life the left coronary being formed before the right The endothelial buds appear in the truncus arteriosus before it is subdivided into aorta and pulmonary artery by the development of the spiral septum Grayzel and Tennant (1934) reporting a case of tricuspid atresia with coronary arteries arising from the pulmonary artery suggest that the anlage of the coronary artery appears in the anterior part of the distal bulbar endocardial cushion destined to form the anterior cusps of the aortic valve If the coronary buds are in the posterior parts of the endocardial cushions the coronary arteries will arise from the pulmonary artery The mechanism is diagrammatically shown in fig 36 It is apparent that displacement or faulty development of the spiral septum or a misplaced bud may result in one or both buds coming within the pulmonary artery It is therefore possible for the following abnormalities to occur The classification of King (1941) has been followed

Abnormalities of Origin

(1) Origin from a common arterial trunk gross abnormalities of the heart transposition

(2) Origin from the pulmonary artery
  (a) Left coronary
  (b) Right coronary
  (c) Both coronary arteries
  (d) Accessory coronary artery
(3) Abnormal origin from a normal aorta
  Single coronary artery
(4) Anomalies of number
(5) Anomalies of distribution
(6) Aneurysm

ORIGIN OF CORONARY ARTERIES FROM A COMMON ARTERIAL TRUNK

ABNORMAL ORIGIN ASSOCIATED WITH MALFORMATION OF HEART

Where there is a single arterial trunk as in cor biloculare or cor triloculare the single vessel gives off two pulmonary branches and then continues as the arch and descending aorta with their appropriate branches There is often a single coronary artery In complete trans position of the vessels both coronaries spring from the transposed aorta and venous ventricle Failure of torsion resulting in partial transposition changes the position of the vessels relative to the aortic cusps (Abbott 1927) In aortic atresia the coronary arteries are found in their usual position but receive their blood through a patent ductus into the cul de sac of the atretic aorta In some complex abnormalities the coronary artery has arisen from the carotid artery (Vernon 1856) or from a pulmonary branch (Dreyfuss 1929 Tow 1931)

Origin from Pulmonary Artery

(1) LEFT CORONARY FROM THE PULMONARY ARTERY There are twenty seven confirmed cases in the literature to date the most recent being those of Carrington and Krumbhaar (1924) Bland White and Garland (1933) Sanes and Kenny (1934) Chown and Schwalm (1936) Barnard (1938) Soloff (1942) Proescher and Baumann (1944) Diaz (1945) Eidlow and Mackenzie (1946) Lyon Johansmann and Dodd (1946) and Kannitz (1947) Soloff has reviewed the existing literature and con siders that the condition is not incompatible with life if there is an adequate coronary circulation He describes a clinical picture in young children (page 96) in whom symptoms may appear in the second or third month and death a few months later The characteristic pathology has been mentioned above and consists of marked degenerative change with fibrosis and occasionally calcification in the left ventricle These changes are identical with those induced by coronary sclerosis in the adult and may be accompanied by electrocardiographic changes with T wave invasion in one or more leads The T wave was inverted in leads 1 and 2 of the case of Bland White and Garland and there was

low voltage T wave inversion was also present in the case of Lyon *et al* (1946) and Kaunitz (1947)

In some cases the abnormality has been compatible with adult life Abbott s (1927) case met an accidental death at the age of 64 The descending branch of the left coronary entered a large sinus which was *in communication with large vessels* Rubberdt s (1937) 27 year old man died suddenly at work and a greatly enlarged right coronary artery anastomosed with the left Kickel s (1934) case aged 38 had also a very large right coronary artery and there were anastomoses between the two coronary vessels Dietrich s (1939) patient a man of 57 had hypertension angina pectoris and auricular fibrillation It is evident that an effective collateral circulation was present in these cases who survived to a later age

(2) Right Coronary from Pulmonary Artery There are but two recorded cases of the abnormality those of Monckeberg (1914) and Schley (1925) Both cases aged respectively 30 and 61 died of inter current disease unrelated to their anomaly There were no changes of note in the right ventricular musculature The right coronary artery was thin walled like a vein and it is assumed from the absence of pathological changes that there was an adequate anastomosis between the two coronary arteries Possibly as mentioned above venous blood contains sufficient oxygen for the lesser metabolic needs of the right ventricle or the Thebesian vessels furnished an adequate collateral circulation

(3) Both Coronary Arteries from Pulmonary Artery This must not be confused with the state found in a complete transposition of the vessels where both coronary arteries are filled from the venous ventricle Only two cases are known those of Grayzel and Tennant (1934) and Limbourg (1937) The former case was accompanied by a gross mal formation with tricuspid atresia and the latter showed left ventricular hypertrophy Both died at the tenth day with cyanosis and extreme dyspnoea

(4) Accessory Coronary Arteries These are generally small and of no clinical significance

Abnormal Origin from a Normal Aorta

The coronary artery especially the right may arise at a higher level than usual above the aortic cusps A coronary artery may also arise at an unusual sinus Thus both coronaries may arise from the same sinus and own the same orifice These cases are often described as having an absent coronary artery or as a single artery This designation may be anatomically correct but is functionally incorrect because the branches of the vessel arising from the other sinus of Valsalva take over the territory of the vessel that has failed at its usual point of origin Jones s (1937) case had a single coronary artery springing from the

right sinus which divided at once into three branches one the right coronary and the other two branches corresponding to the left coronary In Smith and Graber s (1926) cases with coronary thrombosis a single right coronary divided into two branches one corresponding in its distribution to the left coronary Gallavardin and Revault s (1925) case is similar In King s (1940) case a large right coronary artery immediately divided into three branches the main vessel corresponding to the right coronary and most of the circumflex branch of the left coronary The intermediate vessel took the place of the anterior branch of the right coronary and the third branch replaced the anterior descending branch of the left coronary artery

Anomalies of Number

Diminution in number is discussed above Where there is a reduction the other vessel gives branches which take over the distribution of the absent vessel

Three or four vessels may arise from the aorta and an accessory vessel may arise from the pulmonary artery These are without clinical significance

Anomalies of Distribution

There are many minor differences in distribution which can be regarded as anatomical variants of the normal The commonest is when the circumflex artery of the left coronary extends to give a posterior descending branch and supplies an area of posterior aspect of the right ventricle Other anomalies occur in connection with the single or abs nt vessel above

ANEURYSM The first reported case is that of Malet (1887) A cirsoid aneurysm of the right coronary artery was present in the case of Harris (1937) The aneurysm communicated with the right auricle There was a deficiency of elastic tissue in the vessel and it was thought that the connection with the auricle was due to persistence of a foetal communication which is ordinarily reduced to capillary size

An arteriovenous aneurysm of the right coronary artery and coronary sinus has been described by Halpert (1930) in a man of 54 The right coronary artery expanded immediately after its origin into a series of large vascular loops opened by a short vessel into a dilated coronary sinus The condition was symptomless during life Lowenheim (1932) reported an arteriovenous aneurysm of the right coronary artery and vein

Localized dilatations may also occur upon the walls of the coronary arteries especially the left corresponding to those seen in the cerebral vessels and have even been found in the interventricular septum They are liable to rupture causing a haemopericardium and death They are symptomless until some accident occurs and are unlikely to be recognized until post mortem examination

An interesting and unusual case is that of Biorck and Crafoord (1947) where an arteriovenous aneurysm of an aberrant branch of the left coronary artery communicated with the pulmonary artery There was a machinery murmur identical with that of the patent ductus The abnormality was revealed on exploration of the heart and the aneurysm was tied between ligatures with cure of the patient who was aged 15

CLINICAL PICTURE Only those cases where the left coronary artery arises from a pulmonary artery need be considered Reported cases



FIG 37 Radiograph of child aged 2 with gallop rhythm and gross enlargement of the heart The electrocardiogram (fig 38) was abnormal and a diagnosis was made of abnormal origin of the coronary arteries

suggest that the infant appears to be normal at birth but after a few days to months there is evident respiratory distress with dyspnoea cough and vomiting These symptoms are often worse after food has been taken There may be pain of a colicky nature which Soloff believes to be angina pectoris because of the shock like facies pallor and sweating induced by eating Clinical examination reveals an enlarged heart of which confirmation is received from the X ray picture There are usually no murmurs present but a gallop rhythm is common A personal case followed for a period of fifteen years shows at the age of 15 marked impairment of exertional capacity but the general development of the patient is good for his age

RADIOLOGY The heart is grossly enlarged the left ventricle being mainly involved (fig 37) In the left anterior oblique position the left ventricle is enormous and the right ventricle may be slightly enlarged Cardiac calcification in a young subject might be suggestive of this abnormality

ELECTROCARDIOGRAM The electrocardiogram is abnormal and suggestive of the changes of coronary thrombosis There is inversion of the T waves in leads 1 and 2 and often in lead 3 Axis changes are infrequent (fig 38)

DIAGNOSIS The differentiation of this group of cases from other conditions in which the heart is greatly enlarged turns upon the electro cardiographic findings which are characteristic Anomalies which may have to be differentiated are von Gierke s disease rheumatic carditis

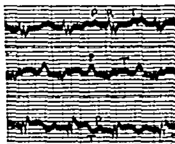


FIG 38 Electrocardiogram of child aged 2 in whom an abnormal origin of the coronary arteries has been diagnosed

severe anaemia and the acute form of myocarditis of which Fielder s myocarditis is the type

PROGNOSIS In general Soloff found that the origin of both coronary arteries from a pulmonary artery lead to death in a few days A left coronary artery arising from a pulmonary artery was compatible with life if there was an adequate collateral circulation Most often the collateral circulation is inadequate and progressive ischaemic necrosis and subsequent fibrosis leads to death below the age of one year A few cases survive to adolescence or adult life and survival has been known to the age of 64 (Abbott)

Abnormalities of the Pulmonary Artery

Absence of a main pulmonary artery is rather rare but it is worthy of consideration because it is an abnormality that may be encountered when operation is performed upon a cyanotic child Blalock has observed absence of a pulmonary artery branch on two occasions in his series

In Fraentzel s (1868) woman of 25 the right branch of the pulmonary artery was absent There was an aortic septal defect of 12 mm diameter between the aorta and pulmonary artery Just below the arch the aorta gave off a large vessel which entered the right hilus The ductus arteriosus and foramen ovale were closed

In Doering s (1914) case the right pulmonary artery was absent In

its place a large vessel was given off the innominate artery The ductus arteriosus was widely patent Ambrus (1936) reported a case in which the right branch of the pulmonary artery was absent and the widely patent ductus arteriosus was continuous with the descending aorta An anomalous right pulmonary artery was given off as a branch of the innominate artery In Muller s (1938) case the right branch was com pletely absent and ischaemic necrosis of the right lung resulted in a pneumothorax on the fourth day of life

In a different category are the cases of Statman and Zawisch (1947) In the first case a pulmonary trunk gave off a right branch and beyond this passed through a widely patent ductus arteriosus into the aorta The innominate and left carotid arose from the ascending aorta The right lung was doubled in size and the left lung existed as a rudiment behind the aortic arch In their second case a truncus aorticus solitarius a pair of pulmonary arteries arose from the back of the descending aorta and were in all probability an adapted pair of first intercostal arteries In other cases of single aortic trunk a patent ductus arteriosus divided into two pulmonary arteries as in the case of Popjak In Shapiro s case a left and right pulmonary artery arose from the aorta about 2 cms above the cusps The right pulmonary artery gave off an innominate artery Anomalous pulmonary vessels may take origin at about the level of the tenth thoracic vertebrae and pass laterally to the lower lobes of the lung Examples are those collected from the literature by McCotter (1910) In addition a pulmonary vessel may arise just at or occasionally just below the level of the diaphragm Lastly an anomalous artery may arise from the coeliac axis (Batts 1939) from the right phrenic artery from the innominate artery or from the right subclavian (Menke 1936) Douglass (1948) points out that a great many cases have been associated with a pulmonary cyst confirming their origin as congenital anomalies

Most of these anomalies are capable of embryological explanation There are six pairs of aortic arches in the embryo not all present at the same time The pulmonary artery trunk develops from the truncus arteriosus and the branches from the sixth aortic arches The right subclavian is derived from the fourth right arch The ventral com munications between the third and fourth arches become the common carotids The communication between the fourth and sixth arches on the right side becomes the innominate artery The fifth arches the last to appear and the most rudimentary join with the sixth arches and should the sixth arch be absent or fail to develop an anomalous pul monary artery may be developed from the fifth arch In regard to cases with agenesis of a lung the pulmonary branch from the sixth arch reaches its respective pulmonary bud at about the 5 mm. stage of the embryo If the pulmonary artery fails to develop growth and development of the lung may also be arrested

Anomalies of the Great Veins

The coronary sinus is derived from the transverse part and left horn of the sinus venosus In early foetal life a left superior vena cava enters the left horn of the sinus The left vena cava gradually atrophies a vestige remaining as the oblique vein of Marshall The pulmonary veins arise as a single trunk on the dorsal wall of the sinus venosus and as this latter structure becomes incorporated in the auricle they come to lie on the posterior wall of the auricle to the left of the left valve of the sinus venosus When the left valve of the sinus fuses with the septum superius to form the interauricular septum the pulmonary veins enter the left auricle Disturbances of or arrest of normal development of these structures results in numerous anomalies many of which are of little clinical importance and most of which are unrecognizable by clinical methods Perhaps the most interesting association with anomalies of the great veins is a defect of the septum secundum Such defects are situated directly beneath the orifice of the superior vena cava which in itself is abnormally placed and looks into the cavities of both auricles A right pulmonary vein entering the superior vena cava or both pulmonary veins entering the right auricle may or may not be associated with a similar defect

PERSISTENT LEFT SUPERIOR VENA CAVA A persistent left superior vena cava may enter the heart through the coronary sinus and represents persistence of the anterior cardinal vein and duct of Cuvier of the early embryo The various types of persistent left superior vena cava have been classified by McCotter (1915) into five groups

(1) Right and left superior vena cava without anastomosis
(2) Right and left superior vena cava with small anastomosis
(3) Right and left superior vena cava with normal anastomosis
(4) Left superior vena cava with absence of right vessel
(5) Left superior vena cava not falling into the above categories

Of these group (1) is the commonest type and is usually associated with other anomalies such as abnormal termination of the pulmonary veins and defects of the interauricular septum Sometimes the left pulmonary vein opens into the left superior vena cava as in the case of Johnston (1915) A persistent left with absence of the right superior vena cava has been described frequently (Dietrich 1913 Schultz 1914 Abbott 1936) The clinical picture is that of the complicating abnormality and a left superior vena cava may occasionally be recognized during life by the radiological picture which shows widening of the superior mediastinum

Anomalies of the Pulmonary Veins

Arrest of development may take place so that both pulmonary veins enter the coronary sinus by a common trunk and thence the right auricle The foramen ovale remains patent as the sole means whereby

blood may enter the left side of the heart and so reach the systemic circulation The cases of Nabarro (1903) and Abbott (1936) both female children aged 5 months and 2 years respectively and the case of Sanes (1939) are examples of this anomaly The pulmonary veins may drain directly into the right auricle as in the nine months old child reported by Duff (1938) In this case the foramen ovale was patent and there was no trace of a valvular flap over the foramen Absence of the valve was interpreted as an indication that the foramen primum had formed the final septum and that the left valve of the sinus venosus had not been incorporated into the auricular septum so that the pulmonary veins remained in the right auricle

The pulmonary veins may enter the superior vena cava Several types of this anomaly have been reported The right pulmonary veins may enter the superior vena cava the left pulmonary veins being normally sited The left pulmonary veins may enter a persistent left superior vena cava or a left innominate vein The entire venous return of the lungs may empty into a left superior vena cava and pass through an innominate vein to reach the right superior vena cava and the right auricle The cases of McCotter (1915) and McManus (1941) are good examples In Gillespie s (1941) case the lungs were joined together by a single vein from the middle of which arose a thick trunk which entered the lower posterior part of the superior vena cava Lastly a number of bizarre cases have been reported in which a pulmonary vein has entered such structures as the inferior vena cava the azygos vein the subclavian vein or the portal vein

PATHOLOGICAL EFFECTS OF PULMONARY VEIN ANOMALIES Anomalies of the pulmonary veins result in some or all of the venous return from the lungs entering the right auricle As a result hypertrophy of the right auricle and ventricle ensues and the pulmonary artery dilates The degree of these changes will vary being less in those cases where part of the venous return is abnormally diverted and extreme where the whole venous return of the lungs is returned to the right auricle The left auricle remains small and the only orifice entering it is the foramen ovale It is through this foramen that the systemic circulation is supplied A certain number of cases are associated with gross defects of the posterior part of the auricular septum In others the limbus of the fossa ovalis may show no valve flap but in a porportion of cases the foramen ovale may be furnished with a valve At autopsy apart from these general findings the liver is enlarged and the lungs congested owing to failure the commonest cause of death

In regard to the circulation it is obvious that a considerable volume of oxygenated blood will be recirculated in the pulmonary circuit The systemic circulation in the extreme forms of the anomaly is solely supplied through the foramen ovale and to this extent the existence of the patient is determined by the degree of patency of this orifice

Cyanosis is only present as a terminal event and is then accompanied by congestive heart failure The cyanosis is mainly due to peripheral stasis

CLINICAL PICTURE Enlargement of the heart is the rule and the left chest bulges The heart may be so enlarged as to appear to occupy the whole left chest There is no cyanosis until failure supervenes and it is a terminal event A systolic murmur may be present or absent and when present is thought to be due to the passage of blood through the foramen ovale The systolic murmur is loudest at the base The pulse is small and the blood pressure low Abnormalities of the cardiac rhythm are common either paroxysmal tachycardia flutter or a persistent sinus tachycardia Later there are the signs of congestive heart failure with enlargement of the liver

RADIOLOGY The heart is enlarged to right and left with a prominent pulmonary conus and congestive changes in the lung fields In those cases where there is a persistent left superior vena cava there is widening of the vascular pedicle due to the shadow cast by the distended anomalous vessel The oesophagus is not displaced The oblique view clearly demonstrates the gross enlargement of the right cavities

ELECTROCARDIOGRAM Even in the more minor degrees of the abnormality there is right axis deviation The arrhythmias indicated above may be present

DIAGNOSIS It might be possible to make a diagnosis during life if the possibility is borne in mind The salient feature is a grossly enlarged heart without cyanosis and with a right axis in the electrocardiogram The newer methods of cardiac catheterization should show an abnormally high oxygen content in samples withdrawn from the right auricle and the figures of oxygen saturation obtained might well be identical with those obtained in a sample of arterial blood

In regard to differential diagnosis the auricular septal defect has to be distinguished This may be a matter of difficulty because the X ray picture is not wholly dissimilar Likewise arrhythmias are prone to occur in the auricular septal defect as in other conditions where there is auricular hypertrophy It may however be said that the auricular septal defect rarely causes symptoms in infancy and enlargement of the heart appears much later than is the case with anomalous pulmonary veins

The only possible treatment of a radical nature would be surgical and this has not yet been attempted Digitalis may be given for the control of failure and arrhythmias may be treated with appropriate remedies

CHAPTER VIII

ABNORMALITIES OF THE SEMILUNAR CUSPS

The aorta and pulmonary artery are formed by the division of the truncus arteriosus The distal bulbar septum divides the lateral pair of the four bulbar endocardial cushions with the result that there are three endocardial cushions in each artery From these develop the aortic and pulmonary cusps by a process of hollowing out Irregularity in the growth of the distal bulbar septum whereby one lateral cushion remains unbisected may account for the formation of only two cusps in the aorta The bicuspid pulmonary valve of the tetralogy of Fallott has been explained by Spitzer (1923) on a phylogenetic ba is (page 259) as due to the persistence of the bicuspid reptilian right aorta Simonds (1923) suggested that fusion of the two halves of the bisected lateral cushions might similarly lead to the formation of bicuspid valves In other cases four cusps may be present in one or other of the arteries The fourth cusp is usually small and has the appearance of having been interpolated and may arise from an abnormal supernumerary endocardial cushion Four cusps are infinitely more common in the pulmonary artery than in the aorta (fig 39)

Fenestration of the Aortic and Pulmonary Cusps

Fenestration of the aortic and pulmonary cusps is not uncommon It occurs in the crocodile and other lower vertebrates It is thought by Foxe (1929) that fenestrations were acquired anomalies their incidence increasing with age Fenestrations above the line of closure of the valve are rarely of significance but cases have been reported where they caused aortic incompetence

Bicuspid Aortic Valve

The association of a bicuspid aortic valve with other congenital anomalies is strong evidence of a congenital etiology Anomalies of the aortic valve cusps are most frequently associated with cardiac abnormalities of the acyanotic type and particularly with coarctation of the aorta The other accompanying acyanotic lesions found are localized defects of the interventricular septum aortic septal defects patent ductus arteriosus subaortic stenosis and anomalies of the aortic arch Abbott (1932) suggests that the frequent association of a bicuspid aortic valve with abnormalities of this type may have a bearing on etiology

There remains a group of cases in which a bicuspid aortic valve exists as a sole abnormality In its simplest form the bicuspid aortic valve exists as two valve cusps without any ridge or raphe More often there exists a conjoined cusp with a ridge or raphe that subdivides it This is particularly the case in the adult and an almost precisely similar appearance may be presented by the acquired form of bicuspid valve which results from inflammatory fusion of adjacent cusps and the ultimate disappearance of the fused portion It is likely that the

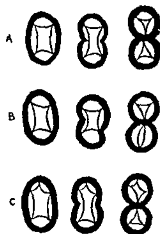


FIG 39 A Normal division of common trunk with two sets tricuspid valve B Abnormal division of common trunk bicuspid aortic valve C interpolated abnormal cushion providing a fourth pulmonary cusp

acquired bicuspid valve is more common than the congenital type now that the two types can be histologically differentiated The frequency of infective endocarditis and the liability of defective tissue to bacterial invasion has led in the past to the assumption that most cases were congenital whereas in fact any deformed valve whatever its etiology is equally susceptible to infection

Because of these varying etiologies the ultimate recognition of a congenital bicuspid valve may depend upon microscopical examination Below the age of five it is safe to assume a congenital origin If there are two large cusps without a raphe or ridge behind one of them or if there is a low thin ridge of healthy appearance the probabilities are that it is a developmental origin Osler (1886) stated that the characteristics of a congenital bicuspid valve were a low almost obliterated raphe a large single cusp with a compensatory shortening of the smaller fused cusp in order to make the valve competent frequent

fusion of the coronary (anterior) cusps and absence of the corpora Arantii These criteria are held to be inadequate by Gross (1937) and Koletsky (1941) Lewis and Grant (1923) found that a low raphe with parallel borders and bifid anterior end was always on microscopical examination of congenital origin This corresponds with the view of Osler

The microscopical examination of the bicuspid valve was first conducted by Lewis and Grant (1923) who made sections of the raphe behind the conjoined cusp Their important findings are concerned with the distribution of the elastic tissue of the aortic media and valve and its relationship to the annulus fibrosus In the normal state a section of the commissure shows that the annulus fibrosus is to be found superficial to the elastic tissue over the top of the commissure as originally described by Monckeberg (1904) A section of the congenital raphe which represents an abortive commissure shows the annulus fibrosus to be deep to the elastic layer which is continuous with that of the subintimal layer of the aorta The congenital raphe thus presents an arrangement of its layers precisely similar to the arrangement prevailing in the middle of a normal cusp and does not show the reversal of the layers normally taking place at the commissure Koletsky (1941) who studied bicuspid valves in the infant in general confirms the histological findings of Lewis and Grant He states that the characteristic appearance of a congenital ridge is that of a long narrow bar like elevation of the aorta uniform in width and depth and running in the long axis of the aorta from the cusp to the sinus of Valsalva

The bicuspid aortic valve is prone to important pathological changes The valve cusps may undergo thickening and fibrosis and ultimately calcification may take place It should not be assumed that many cases of calcific aortic stenosis are of congenital origin The recent monograph of Karsner and Koletsky (1947) suggests that most cases are rheumatic in origin The results of such processes are scarring and deformity of the cusps with the development of aortic stenosis and incompetence and later failure of a progressive type There is a liability to sudden death either by rupture of a dilated ascending aorta or by rupture of an aneurysm developing in the wall behind the abnormal cusp Death may also occur suddenly from syncope in cases where there is extensive calcification Even in the relatively healthy bicuspid valve strain may provoke incompetence and failure By far the most important complication of the bicuspid valve congenital or acquired is infective endocarditis The liability of these structures to infective process was recognized by Graves (1843) and was stressed by Osler (1886) Lewis and Grant (1923) and Grant Wood and Jones (1928) showed that infection of the valve was not due to embolic processes but was due to the arrest of germs circulating in the blood stream in parts of the valve

weakened by sclerosis and degenerative changes Ulcerative and destructive processes are set up by organisms that are not necessarily especially virulent the commonest being the *streptococcus viridans* The appearances in infective endocarditis are striking and the luxuriance of the vegetations is out of all proportion to the virulence of the infecting organism The vegetations may spread to the aortic conus and on to the anterior cusp of the mitral valve

CLINICAL PICTURE There are no signs or symptoms which permit a diagnosis Diagnosis if attempted is purely inferential and is permissible when aortic incompetence is found in the presence of lesions known to be associated with a bicuspid aortic valve such as coarctation of the aorta The lesion may also be suspected when there is an infective endocarditis of the aortic valve in the absence of rheumatic history

COURSE AND PROGNOSIS The subject may lead an ordinary normal life without untoward incident As indicated above there is a risk of sudden death By far the greatest hazard is infective endocarditis and this occurred in 20 *per cent* of Abbott s series The average age at death is 33 years (Abbott)

Quadricuspid Aortic Valve

An aortic valve with four cusps with aortic incompetence was reported by Froment Bertoye and Perreau (1947) Such cases are rare and supernumerary cusps do not appear to have the liability to bacterial invasion presented by the bicuspid valve Wauchope (1928) found no example of a quadricuspid valve in 1 966 autopsies

Pulmonary Valve

A bicuspid pulmonary valve as a sole abnormality is a very rare lesion It is the constant accompaniment of the tetralogy of Fallot or cases with dextroposition of the aorta or other grave defect and is held by Spitzer to represent a persistent right (reptilian) aorta A supernumerary pulmonary cusp occurs four times in the series of Wauchope Five cusps are figured in a case of Peacock Such supernumerary cusps are often smaller than normal cusps

Abnormalities of Septation

ANOMALOUS SEPTA IN THE LEFT AURICLE This condition sometimes referred to as a double left auricle is rare Palmer (1930) collected ten cases and added a further example A similar case has been reported by McLester Bush and DuBois (1940) The left auricle is subdivided into two cavities an anterior lower and an upper posterior by a thin perforated diaphragm which rises from the wall of the left auricle and joins the interauricular septum above the open or closed foramen ovale The anterior lower cavity contains the mitral orifice while the posterior upper cavity receives the pulmonary veins The two cavities

communicate by slit like perforations and the diaphragm has the same effects as a mitral stenosis

The abnormality probably arises secondary to a misplaced pulmonary vein This leads to displacement of the septum primum to the left the vein intervening between the septum primum and secondum Palmer suggests that the upper cavity represents the dilated end of the foetal pulmonary vein and is therefore not truly auricular The lower cavity would then be wholly auricular and the observation of McLester *et al* that the diaphragm contained muscular tissue would support this view

There are no physical signs which permit recognition of the anomaly during life

ANOMALOUS SEPTA IN THE RIGHT AURICLE Chiari (1897) described network of fibres in the right auricle lying between the interauricular septum and the Eustachian and Thebesian valves guarding the orifices of the inferior vena cava and coronary sinus Generally the network amounts to a few fibres but it may assume the proportions and importance of a septum It is probably the vestige of the right venous valve The importance of the condition lies in the liability of the network to become the seat of a thrombus formation with its subsequent risks of pulmonary embolism This caused death in a case of Chiari Curiously enough the network sometimes serves to arrest a thrombus which might otherwise have caused death from embolism (Haas 1916) The condition which is present in 2 3 per cent of otherwise normal hearts (Yater 1936) usually gives rise to no definite signs or symptoms A continuous humming murmur with systolic accentuation was present in the case of Wilson (1938) A large anomalous septum in the case of DuBois and Hollinshead (1944) probably represents a persistent right valve of the sinus venosus Free muscle cords in the auricles have been described by Brenner (1938)

ANOMALOUS SEPTA IN THE VENTRICLES The right ventricle may be divided into two chambers in conus stenosis of the pulmonary artery This arises as an abnormality of involution of the bulbus cordis the orifice in the septum dividing the ventricle being the lower bulbar orifice or ostium infundibuli Such cases have been reported as cor triventriculare and are referred to on page 179

A second group of cases is where a rudimentary ventricle is cut off from a common ventricle into which both auriculoventricular orifices open The transposed aorta generally arises from the rudimentary anterior ventricle (fig 90) These cases are more fully discussed under triloculate hearts

ANOMALOUS CHORDAE These are of no clinical importance but may stretch across the cavity of the left ventricle They may give rise to peculiar auscultatory signs In the case of Hamilton (1899) a murmur was audible fifteen feet from the patient Congenital cord like structures stretching across the aortic orifice have been described by Archer

(1878) Rizzi (1935 1939) and Brody (1942) Their exact origin is unknown They extend from above one commissure to an adjacent commissure

Congenital Arteriovenous Aneurysm

This rather rare condition is the result of a communication between an artery and a vein Hunter (1757) probably the first observer of an arteriovenous aneurysm thought the condition was always traumatic in origin Hewitt (1867) reported a congenital arteriovenous aneurysm involving the right iliac vessels Halsted (1919) considered that con genital aneurysms were very rare and were slightly more common when associated with a naevus than when such an association was lacking Rienhoff (1924) was able to collect twenty two cases to which he thought a definite congenital etiology could be assigned and he further noted the close association between haemangiomata and congenital arteriovenous fistulae Pemberton and Saint (1928) state that of twenty five patients with arteriovenous aneurysm operated on in the Mayo Clinic between 1915 and 1936 sixteen were acquired and nine congenital in origin

The difficulties in distinction between the acquired and congenital forms are enhanced by the apparent latency of some congenital cases until symptoms are evoked by strain There is an etiological relation ship between the angiomata capillary naevi and arteriovenous aneurysms for trauma in any of these conditions may lead to an arteriovenous communication Similarly related is the congenital phleboarteriectasis with associated haemangiectatic hypertrophy of a limb of Parkes Weber (1918) Klippel and Trenauny had previously described in 1900 a syndrome characterized by cutaneous naevus congenital varicose veins and hypertrophy of bones and tissues of the involved limb Most commonly a leg is involved but an arm could be similarly affected The varicosities are not always apparent in the very young and tend to appear at puberty This syndrome is but a part of the more diverse haemangiectatic condition described by Parkes Weber and the term Parkes Weber Klippel syndrome has been applied to those cases in which hypertrophy of a limb has been associated with naevus and enlargement of arteries or veins or both Increase in the length of the bones of the affected limb and advancement in time of the centres of ossification has been thought due to increased arterial blood supply and possibly to an increased size of the nutrient vessel Servelle (1947) considers that venous stasis is the more important factor in bone growth and suggests that lumbar sympathectomy is appropriate for the relief of the condition The point may be made that this anomaly should be considered as a potential aneurysm under the influence of trauma or in states where the blood flow is increased from any cause or if there is obstruction to the venous return

Rienhoff (1924) from a study of the pig embryo has suggested that the lesion is the result of persistence of vessels or communications of the primary anlage which failing to develop into the normal definitive vascular tree results in the formation of anastomotic channels between the otherwise normally developed arterial and venous trunks It is thus remarkable that congenital arteriovenous aneurysms are not more frequent in view of the common origin of both sides of the vascular tree The acceptance of this view that arteries and veins arise through differentiation from a common capillary plexus (Pemberton and Saint 1928 Seager 1938) reconciles the essential basal similarity of the above lesions This view is endorsed by Dean Lewis (1930) who also remarks upon the latency of the condition and its recognition at about the age of puberty when strain may open up the channels and cause an actual arteriovenous aneurysm of clinical significance The extent of such a communication may be recognized by arteriography and venography An important arteriovenous fistula developing before puberty is generally accompanied by changes in bone growth (haemangiectatic hypertrophy) Aneurysms developing after the closure of the primary epiphyses are unaccompanied by such changes They however may provoke important changes in the heart which may become greatly enlarged in certain cases Rienhoff (1924) and Abbott (1927) have stated that enlargement of the heart is absent in the congenital arteriovenous aneurysm although such hypertrophy of the heart was present in the cases of Nicoladini (1875) and Israels (1877) It is remarkable that a comparable fistula such as the patent ductus arteriosus does not result in the degree of cardiac hypertrophy occasioned by a peripheral arteriovenous fistula This is because vasoconstriction in the smaller pulmonary branches limits the blood flow through the ductus into the pulmonary artery by maintaining pressure in the pulmonary artery In the more peripheral fistula blood escapes readily into the low pressure venous system against little resistance

An arteriovenous aneurysm may occur in almost any situation but is particularly associated with the limbs It may involve the external carotid artery and external jugular vein as in the cases of Rienhoff and Bigger and Lippert (1937) The ear may be involved as in the cases of Eve (1880) and Lewis A unique case has been described by Halpert (1930) where such an aneurysm formed between the right coronary artery and the coronary sinus The essential anatomical feature is a communication between an artery and vein without the intervention of a capillary bed The artery is greatly dilated proximal to the lesion and reduced in size beyond it The veins are dilated and varicose and oedema of the affected limb may be present As stated above enlargement of the heart is present in a limited number of cases and is of interest because cure of the aneurysm by surgical intervention may lead to a return of the heart to a normal size This is especially well

shown in the case of Quattlebaum (1937) Various explanations have been offered of the cardiac hypertrophy Holman (1923) suggests the increased work by the heart owing to an increased venous pressure and volume flow through the heart leads to its hypertrophy and dilatation On the other hand Lewis and Drury (1923) do not admit any rise in venous pressure other than that resulting from congestive failure and state that the changes in the heart are the result of a diminished coronary supply and poor nutrition of the heart Whilst all the factors may not as yet be known it does seem certain that the diversion of blood into the venous system implies an increased venous return to the heart The heart responds to this by dilatation and later by hypertrophy because of the increased output that it has to maintain The greater the output the greater the chances of hypertrophy and the presence of symptoms and signs of heart failure This process is reversible and surgical cure of the fistula leads to a return of the heart to a normal size Where the condition has been present for a long time the enlargement is more slowly reversible

CLINICAL PICTURE In its earliest stages the presence of a fistula may be revealed in young subjects by an indolent ulcer associated with a naevus varicose veins or other vascular lesion and slight hypertrophy of the affected limb In the well developed case an expansile tumour is present This becomes larger as the lesion progresses perhaps in response to trauma or strain A systolic or continuous thrill is palpable over the mass and corresponding systolic murmurs or continuous hums may be present Where the lesion involves a limb the veins are markedly varicose and may pulsate Considerable oedema is present There may be trophic changes in the skin and indolent ulceration due to short circuiting of the blood supply A slightly increased temperature may be present in the affected part In cases where the lesion has been established since an early age excessive growth of the limb or neighbouring bones may be present and is thus an indication of the age of the lesion Aneurysms developing after puberty are unaccompanied by changes in bone growth Gilmour and Bolam (1937) have suggested that such changes are due to excessive hyperaemia and the added factor of a raised local temperature Others have attributed bone growth to venous stasis Pressure on the artery supplying the aneurysm leads in most cases to a transitory rise in the blood pressure and slowing of the pulse and bradycardiac phenomenon of Branham (1890) This sign is usually less marked in the congenital case Lewis and Drury (1923) consider this to be a vagal effect As stated above it is exceptional for the heart to be enlarged in the congenital type but hypertrophy appears to be common in the acquired type

COURSE AND PROGNOSIS This will to some degree depend upon the site and extent of the lesion If for any reason the case remains untreated and many cases are amenable to ... of intervention the

tendency is towards the development of congestive heart failure The average age at death is given by Abbott (1931) as 21 with an extreme of 35 years

TREATMENT This may be difficult owing to the actual number of arteriovenous communications present If the communications are few as demonstrated by arteriography the fistulae can be closed by surgical intervention Ligature of the artery proximal to the aneurysm or ligature of the artery and its accompanying veins may not always succeed and amputation may be necessary later on account of gan grene or other complication Lewis s (1930) collected cases indicate that amputation was necessary in thirty four cases subsequent to ligation Other methods of treatment are by injection of the veins with sclerosing solutions compression of the varicosities by rubber bandages and perhaps the application of radium In the acquired case the traumatic arteriovenous aneurysm treatment is more successful and is by excision of the involved area preferably three to six months after injury when an adequate collateral circulation has developed

CHAPTER IX

CYANOSE TARDIVE, PATENT DUCTUS ARTERIOSUS

The characteristic feature of the lesions of the group cyanose tardive is that there exists a communication between the two sides of the heart Such a feature is present in uncomplicated cases of aortic septal defect defects of the auricular and ventricular septums and in the patent ductus arteriosus Under ordinary conditions the pressure in the left side of the heart is higher than that in the right side and consequently any shunt that may be present is from the left to the right or arterio-venous (fig 40) That such a shunt does take place in the anomalies of this group is betrayed by the anatomical changes that take place when an infective endocarditis is superimposed or by the presence of a dilated pulmonary artery in the patent ductus and in the inter auricular septal defect If for any reason such as pulmonary infection or as a terminal event the pressure in the right side of the heart comes to exceed that in the left the shunt is often reversed and becomes a venous arterial shunt This transient or permanent reversal of flow or shunt gives rise to either transient or permanent cyanosis and was first described by Bard and Curtillet (1889) as cyanose tardive

The clinical features of the group as a whole are somewhat similar to those of the acyanotic group and were it not for the possible inter vention of a transient or terminal cyanosis these cases might justifiably be classified as acyanotic With the possible exception of the inter auricular septal defect the actual development of cyanosis is uncommon clinically and its significance rarely recognized The interauricular septal defect presents certain other peculiarities for cases with this lesion rarely if ever develop infective endocarditis but are peculiarly prone to acquired valvular disease Coarse thrills and murmurs are characteristic of the group with the exception of the auricular septal defect Symptoms are rare The contrasting marked physical signs and absence of symptoms usually lead to the patient seeking advice when complications have developed Alternatively the lesion may be dis covered at routine examination and the marked physical signs may lead to unnecessary restrictions being imposed The frequency with which an interventricular septal defect is associated with other cardiac anomalies has raised doubt in the minds of some (Laubry and Pezzi 1923) as to whether it exists commonly as a sole abnormality Those engaged in the routine examination of school children will suspect that it is among the most frequent of all abnormalities The patent ductus

arteriosus not only has a predilection for the female but above all other lesions it is peculiarly susceptible to infective endocarditis It is the only curable lesion in the group

Patent Ductus Arteriosus

HISTORICAL NOTE The ductus arteriosus was known to Galen as a *small vessel* uniting the aorta and pulmonary artery and he was aware of its closure after birth Botallus (1564) never described the ductus to which his name has been attached (Ductus Botallo) but actually described post natal patency of the foramen ovale Carcano (1574)

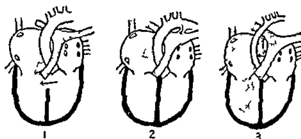


FIG 40 The circulation in the group with cyanose tardive 1 Maladie de Roger 2 patent ductus arteriosus 3 interauricular septal defect The normal tendency of a shunt is from left to right

gives a detailed account of the ductus Laurens (1599) gives pictures of the ductus and foramen ovale which are reversed and anatomically incorrect Harvey (1628) having described the foramen ovale speaks of the ductus in these terms The other union is of the artery like vein (which occurs after that vein has left the right ventricle and is dividing into two branches) It is a sort of third trunk added to these two an artery like channel so to speak leading obliquely from this point to the great *artery and perforating into it* *This channel in the adult* narrows and dwindles Finally it dries up and ceases to exist Senac (1749) was aware of persistent patency of the ductus and thought that this occurred in cases where there was difficulty in breathing Meckel (1812) was surprised to find the ductus rarely open and assumed that an obstacle to the pulmonary circulation was a primary cause of patency Babington (1847) showed a specimen to the Pathological Society of London from a patient of Addison mentioning that Wilkinson King had *correctly diagnosticated patesence of the ductus* arteriosus Bernutz diagnosed a patent ductus in 1849 Peacock (1858) thought that a patent ductus was commonly combined with obliteration of the pulmonary artery and was there to assure a pulmonary circulation In other cases where there was no pulmonary stenosis it was *attributed* to maldevelopment of the aortic arches Turner (1862) considered that a patent ductus was due to an anomaly of the sixth left arch Langer (1857) appears to have given the first account of the

histological changes in closure of the ductus Gerhardt (1867) investigated the physical signs Walkoff (1869) considered that changes in the position of the ductus whereby it became kinked at the pulmonary end at the onset of pulmonary respiration was an added factor in closure Barnard (1939) described pathological changes in the ductus wall during the course of involution The functions of the ductus have been further elucidated by Barclay and his colleagues (1944) The first description of the pathognomonic machinery bruit is that of Gibson (1898) Operation and ligature was proposed by Munro (1907) and first successfully effected by Gross (1939)

ANATOMY In foetal life the ductus arteriosus is a short vessel about 1 cm long and of 5 mm or more in diameter It intervenes between the aorta and pulmonary artery joining the former at a point just distal to the origin of the left subclavian artery It is often a larger channel than is generally realized being equal in calibre to the arch of the aorta or even the pulmonary artery (Harvey 1628 Noback and Rehman 1941) Gerhardt (1867) classified the ductus into several anatomical types He described the cylindrical the funnel shaped with apex towards the pulmonary artery the window type consisting of a direct opening between aorta and pulmonary artery and an aneurysmal type It is so far impossible to identify these types during life or from clinical signs but perhaps angiocardiography may be useful at some future date The practical point is that the window type is not so readily amenable to surgery as are the other types Keys and Shapiro (1943) found ten such examples in sixty cases Crafoord (1944) has successfully operated upon this type of case The ductus whatever its anatomical type may be brittle and friable because of degenerative changes in its walls

The ductus arteriosus may remain as a sole abnormality or it may persist as a complication of or in compensation of other anomalies

An aortic septal defect is to be distinguished by its position relative to the left subclavian artery It may give rise to clinical signs identical with the patent ductus

ROLE OF THE DUCTUS ARTERIOSUS IN FOETAL LIFE In embryonic life the pressure in the right side of the heart exceeds that in the left In the foetus oxygenated blood from the placenta together with venous blood from the superior and inferior vena cavae enters the right auricle This mixed blood is then diverted partially through the foramen ovale to reach the left auricle and ventricle, and thence the aorta and the vessels of the head and neck The remainder of the blood entering the right auricle passes to the right ventricle and traversing the pulmonary *artery and patent ductus arteriosus reaches the descending aorta and* thence the trunk and lower limbs For all practical purposes only a very small volume of blood passes through the lungs in intrauterine life After birth with functional employment of the lungs pressure in

the right side of the heart becomes less than that in the left The ductus then undergoes involution so that by the third week it is almost entirely closed Final obliteration usually takes place in the first three months of post natal life but may be deferred to a much later date The brilliant work of Barclay Franklin and Pritchard (1944) has illuminated our knowledge by radiographic methods

Closure of the Ductus and Pathogenesis of Persistent Patency

Functional closure of the ductus may be completed in less than a minute (Barclay *et al* 1938) The exact changes which make this possible have given rise to much speculation and discussion There has been no uniformity of opinion as to the exact histological structure of the ductus or in regard to the changes that take place in it during its conversion into the ligamentum arteriosus Undoubtedly some divergencies of histological opinion may perhaps be explained by the varying time of its closure Christie (1930) found 65 per cent patent at the end of two weeks and 1 per cent at the end of the first year Wells (1908) and Abbott (1927) have discussed the various theories of closure and generally speaking it seems that this is effected by the combined factors of its own peculiar structure and the pressure changes that come into operation after birth

Microscopical examination of the ductus shows that its structure differs from that of the aorta and pulmonary artery for whereas in these latter vessels the media is largely elastic tissue the media of the ductus is characterized by large amounts of muscular tissue and von Hayek (1935) describes a helical arrangement of the muscle fibres Barnard (1939) has shown that by the fifth month of intrauterine life isolated plaques of intimal hypertrophy develop within the ductus These plaques do not completely encircle the lumen nor do they extend the whole length of the vessel They increase in size until delivery and tend to diminish the lumen In the new born child Barnard finds the inner surface of the ductus lined by longitudinal ridges and the extremities appear more tightly closed than the rest of the vessel On section muscle and elastic bundles in media and intima are separated by a structureless material staining pale purple with haematoxylin At the junctions with the aorta and pulmonary artery there is a clear demarcation as these latter vessels do not present a corresponding change Swensson (1939) and Jager and Wollerman (1942) report somewhat similar findings The subsequent changes which extend over months are of replacement fibrosis and the structureless purple staining material comes to resemble atypical cartilage and collagen fibres appear Barnard finds occasional patchy impregnation with calcium salts In the majority of cases a potential lumen represented by a narrow cleft was observed by Barnard even at the age of 83

In some cases closure of the ductus is accompanied by thrombosis

of its lumen or with a clot at either end of the vessel Portions of such a clot may break off and give rise to embolism in either the pulmonary or systemic circulations A clot at the aortic orifice of the ductus may enter the left subclavian or left common carotid arteries but more commonly it reaches a vessel in the leg A small embolus in a peripheral vessel can cause extensive damage because of the secondary thrombosis which occurs above and below the site of lodgement Thrombosis in the ductus has been described several times since it was first reported by Rauchfuss (1859) Gross (1945) mentions two cases in which there was embolization of and thrombosis in the abdominal aorta It nearly always occurs in the neonatal period at a time when closure of the ductus normally takes place It has been described by Jager (1940) at 55 years and by Altschule (1937) in a patient of 57 The actual clinical condition of a thrombosed ductus may be unsuspected because the physical signs of a patent ductus are lacking in the thrombosed vessel

When the lungs become a functioning mechanism at birth there is a general fall in pressure in the circulatory systems due to the opening up of the vast capillary bed of the lungs and the removal of the placental circulation It has been suggested that the pressures in the right and left sides become more or less equal and under such circumstances no shunt would take place and consequently an obliterative process would be favoured Others have considered the main factors in closure to be mechanical and related to a rise in pressure in the left side of the heart exceeding that in the right This may be effective in two ways either dilatation of the foetal aortic isthmus in response to aortic pressure may exert traction on the aortic end of the ductus facilitating its closure or the aortic end of the ductus may become functionally closed by Strassman s fold a fold of the aortic wall about the mouth of the ductus present at about the seventh month onwards Increased pressure in the aorta would cause this fold to lie across the mouth of the ductus It is very difficult to assess the exact role of any of these factors and probably all of them may contribute in some measure to the final closure of the ductus

In the light of the above it becomes evident that any abnormality of the ductus wall or abnormality of pressure relations after birth may result in persistence of the ductus in its foetal form Atelectasis causing a raised pressure in the pulmonary artery after birth and perhaps at the same time inhibiting the normal dislocation of the thoracic organs may favour the persistence of the foetal state and may figure in the history of the subject Not infrequently abnormality of the aortic isthmus a slight degree of coarctation may be found at post mortem In other cardiac anomalies the ductus may remain patent to assure a circulation to the lungs as in the valvular atresias Again it may remain patent in pulmonary stenosis or other abnormality favouring a rise in pressure in the right heart Lastly abnormalities of the germ plasm

may have some influence and the occurrence of a patent ductus in several members of the same family would lend colour to such a contention Its occurrence in twins has been noted by Smith (1929) and in sisters by Jewsbury (1912) and Brown (unpublished) (figs 42 and 43)

PATHOLOGICAL PHYSIOLOGY A persistent patency of the ductus arteriosus implies a shunt of blood often very large from the aorta to the pulmonary artery and thence via the lungs to the left ventricle to complete an entirely valueless short circuit That the shunt is from left



FIG 41 Patent ductus arteriosus Female aged 3 Note the slight degree of isthmus stenosis of the aorta

to right is shown by the almost constant dilatation of the pulmonary artery and left ventricular hypertrophy of some degree When infection occurs it is the pulmonary artery end of the ductus that is first involved together with the wall of the pulmonary artery opposite the orifice of the ductus suggesting that these are the real sites of strain A large shunt from the aorta results in a low diastolic pressure and is not only a cause of increased work by the left ventricle but also adds to the work of the right ventricle Eppinger Burwell and Cross (1941) have shown that there is a more than 50 per cent increase in left ventricular output The pulmonary artery and its branches dilate and under filling of the systemic arteries is said to cause the slender gracile build and subnutrition that has been so frequently described It has been shown that the volume of the shunt through the ductus may be up to 50 per cent of the output of the left ventricle This means that the pulmonary artery receives not only venous blood on its way to the lungs but also the large additional volume shunted from the aorta The left ventricle is therefore called upon to deal with the added volume of the shunted blood Despite this hypertrophy of the ventricle is rarely a very conspicuous feature although left axis is met with in a small proportion of electrocardiograms Gilchrist (1945) has contrasted the patent ductus with the peripheral arteriovenous fistula which inevitably leads

to cardiac hypertrophy He points out that in the patent ductus constriction of the smaller pulmonary vessels creates an area of peripheral resistance which limits flow through the ductus In contrast in the peripheral arteriovenous aneurysm the blood readily escapes into a capacious venous system at a low pressure The smaller pulmonary vessels tend thus to protect the left ventricle from overstrain In this general connection reference may be made to the work of Lewis and Drury (1923) Keys and Friedell (1939) Holman (1923 1937 1940) and Eppinger Burwell and Gross (1941)

The influence of the volume of the shunt and its effect upon the size of the heart are well shown in the case of identical twin girls (fig 42) The children are 10 years of age at the time of the photographs and the girl with the smaller shunt is three inches taller and more robust than her twin sister

INCIDENCE Patent ductus arteriosus as a sole abnormality is not uncommon and many cases have been brought to light by the routine school medical examinations There is a definite preponderance of female cases In the sixty cases collected by Shapiro and Keys (1943) there were forty six females and fourteen males East (1945) nine females and four males Hunter (1945) ten females and four males Benn (1947) reports thirty three females and thirteen males The series of Brown and Muir of 156 cases shows 108 females and 48 males There is no explanation of this sex incidence in the female Equally the adult case of patent ductus arteriosus is rarely seen at examinations or in the post mortem room It seems possible that the ductus may close spontaneously in the second decade or else the physical signs when failure or other complication occurs may mask the true nature of the case

CLINICAL PICTURE It has been stated that the subjects of a patent ductus arteriosus are of the tall slender type pale and presenting the stigmata of aortic hypoplasia and arterial depletion There is some evidence that where the shunt is very large development may be retarded and this was well exemplified in a personal observation of identical twins (fig 42) who at the age of 10 had classical signs of a patent ductus One child was several inches shorter than the other and its heart was considerably larger indicating a very large shunt Muir and Brown (1932) found that 20 per cent of children with a patent ductus were slightly below the average on comparison with the standard anthropometric tables of their locality This has been confirmed by Gilchrist (1945) who finds under development the exception rather than the rule Cyanosis is absent at rest but it may appear on exertion or with pulmonary infection or as a terminal event In infants it is induced by suckling or emotional experiences and such cases have been mentioned by Smith (1929) and Leech (1932) Permanent cyanosis never occurs and its presence at once invalidates a diagnosis of a



(*a*) (*b*)

Fig 42 Identical twin sisters with a patent ductus arteriosus at the age of 10 The larger heart corresponds to the *smaller twin* This child at the age of 14 developed bacterial endocarditis At operation the ductus was one centimetre in diameter The child made a complete recovery

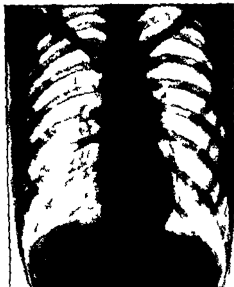




FIG 43 Patent ductus arteriosus in sisters aged 15 and 18 years

ductus as a sole abnormality Epileptiform convulsions may exceptionally occur

Symptoms are occasionally present and are usually those of dyspnoea on exertion fatigue or palpitation Occasionally *dyspnoea may be so* habitual to the patient that he is hardly aware of it and only on operation *realizes how breathless he used to be* A strong functional element may pervade these patients as in the case of the isolated interventricular septal defect The striking physical signs leading to early restriction of

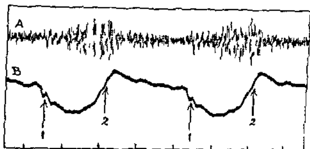


FIG 44 Murmur of patent ductus arteriosus
A stethoscopic phonocardiograph with microphone at site of maximal murmur B *apex cardiograph 1 First heart sound 2 second heart sound*
Typical machinery murmur maximal in the second space at the left sternal edge Stethoscopic phonocardiograph shows the characteristic increase of *intensity of the murmur during systole and the decrease at the time of the* second heart sound
(The arrow marking the second heart sound on the apex cardiograph is *interpolated from reference to another stethoscopic tracing in which there* was less low frequency cut and which showed the second sound well Time intervals 0 2 secs) (Dr M Matthew s case)

activity and possible exclusion from school almost inevitably contribute *to the subsequent development of a neurosis The physical signs may* vary according to the length of the ductus its calibre and the shape of its orifice A small orifice and ductus is likely to give rise to more striking auscultatory signs and conversely a large ductus with concomitant large shunt furnishes a more striking X ray picture The generally accepted pathognomonic murmur of the patent ductus was described by Gibson (1898 1900) and again in 1906 in these words It begins after the commencement of the first sound It is continued during the latter part of that sound and the whole of the short pause It persists through the second sound and dies away gradually during the long pause The murmur is rough and thrilling It begins softly and increases in intensity so as to reach its *acme just about or immediately* after the incidence of the second sound and from that point gradually wanes *till its termination The second sound* can be heard to be loud and clanging This has been confirmed by phonocardiagraphic methods by Routier (1937) and others (fig 44) *The murmur is com*

lesignated a machinery or continuous murmur The murmur is ard in the second left space close to the sternum but in a pro of cases just below the sternal end of the left clavicle Some is the pulmonary artery enlarges more and more to the left the r can be heard at a distance from the sternum in the second left The point to be made is that a machinery bruit is a sharply d bruit but the systolic accentuation may be heard over a much area even over the third and fourth thoracic spines posteriorly st (1945) states that the characteristic machinery murmur may rd in the second and third left spaces a full hand s breadth from rnum in cases where there is great dilatation of the pulmonary due to infective endocarditis Sometimes a machinery murmur difficult to hear and must be very carefully sought with the recumbent and at the end of expiration The murmur may vary deal with the age of the patient In the first two or three years nay be a systolic bruit only loudest over the upper intercostal and transmitted along the left subclavian Usually by the age of r five the murmur has become definitely continuous in quality ossible to detect dilatation of the pulmonary artery radiologically the appearance of the characteristic bruit Crafoord (1944) estioned the specificity of a machinery bruit and mentions two n which a ductus has been diagnosed on the basis of this type of ar but exploration with a view to ligature has disclosed a per truncus arteriosus communis In these cases the maximum ty of the continuous murmur was at the third intercostal space and over the first intercostal space in the other that is to say isual sites

nother case which was explored by Biorck and Crafoord (1947) was an arteriovenous aneurysm formed by an anomalous branch left coronary artery entering the pulmonary artery

doubtedly the patent ductus can be present in the absence of a nery murmur and a systolic murmur characterizes the earlier ed cases for example Gerhardt (1867) Simmons (1906) nter (1903) and later still those of Wessler and Bass (1913) (1931) and Chapman and Robbins (1944) In some cases a ur may be entirely absent and this may be due to either the ce of congesti e failure bacterial endocarditis with blocking of men of the ductus or thrombosis in the ductus

itly a machinery murmur which has been present for years may s and disappear This has happened in five cases in a personal of Brown and Muir and in one of these cases the bruit dis red after seventeen years of observation at the age of 34 This ent spontaneous closure has been noted by others Gross (1939) ked upon it occurrence Shapiro and Keys (1943) and Gilchrist) also mention similar cases Benn (1947) reports two cases in

which a machinery bruit had regressed by the gradual stages of a loud systolic bruit to no murmur at all at the ages of 25 and 27 after observation since childhood

The pulmonary second sound is accentuated and may be well heard but it may be lost in the intensity of the machinery murmur unless it is looked for in the third left space A thrill systolic or continuous in time may accompany a machinery murmur Lastly a distinct diastolic murmur may occasionally be heard along the left *sternal margin and* at the apex This may be due to increased flow through the mitral valve and a relative mitral stenosis It has been attributed to relative pul monary incompetence but it is only right to say that often a case which *shows the maximum of pulmonary dilatation shows no diastolic bruit* The clinical point that cannot be too highly emphasized is that diag nosis is chiefly made on the basis of the machinery murmur and the ready recognition of this is essential if a case is to be treated surgically Gerhardt s ribbon dullness along the left margin of the sternum was at one time considered to be a capital point in clinical diagnosis As is due to a dilated pulmonary artery and as such dilatation can occ in other conditions such as the interatrial septal defect or even pulmonary stenosis its recognition is of little but academic importanc

Other peripheral signs are of interest There may be vigorous puls tion in the neck if the shunt through the ductus is large The pulses ma be unequal the left being larger than the right A difference in amplituc between the femoral and radial pulses has been emphasized by Laubr (1930) and may be conceivably due to slight coarctation which is not uncommon with a patent ductus This has been confirmed by Abbott (1927) and may have etiological significance The pulsus paradoxus has been described but is rarely found The diastolic blood pressure tends to be low and the systolic pressure about normal for the age of the patient The pulse pressure is therefore often increased The low *diastolic pressure* may be correlated with the size of the ductus and shunt With exercise there is a drop in the diastolic pressure for a minute or two and the patient may show swinging carotids a col lapsing pulse and even capillary pulsation Bohn (1938) has insisted that *this fall in diastolic pressure may be an important diagnostic point* and in a young subject with a basal systolic bruit and an accentuated second pulmonary sound a reading of the blood pressure after exercise should always be taken Paralysis of the left vocal cord is a rare compli cation and is due to pressure on *the left recurrent laryngeal nerve*

The Infected Ductus

There is however a great liability to infective endocarditis Libman (1910) considered that a *patent ductus was the most important con* genital cardiac defect *related to* infective endocarditis This peculiar liability has been confirmed by Hamilton and Abbott (1914) and by

Boldero and Bedford (1924) The common infecting organism is the *Streptococcus viridans* Vegetations are found filling the ductus itself upon the wall of the pulmonary artery adjacent to or opposite its orifice and on the aortic cusps from whence infection may spread to the mitral valve as in Perry s (1933) case Rarely in cases where the shunt has been in the reverse direction vegetations may be present on the aortic wall opposite the ductus (Foulis 1884 Rickards 1889) The pulmonary valve usually escapes although it was slightly involved in the case of Bedford and Boldero Hamilton and Abbott were the first to show that infective process began by invasion of the intima at a site of strain and were able to demonstrate that the oldest lesion was at the pulmonary end of the ductus

The infected ductus is an exceedingly grave condition and its recog nition is based upon the signs and symptoms of a septicaemia the local cardiac signs of a patent ductus and the evidence of pulmonary infarcts often multiple which are due to emboli shed from the vegetations in the ductus and pulmonary artery Gilchrist (1945) believes that there is often a characteristic radiological picture and serial films exhibit increasing prominence of the pulmonary artery progressive cardiac enlargement occurring rather rapidly together with patchy con solidation in the lung fields of variable extent and in variable stages of development or regression A grossly enlarged heart in a patent ductus suggests the possibility of infection Pyrexia and other systemic dis turbances are commonly present Confirmation of infection may be secured by blood culture and the other characteristic features of an ulcerative endocarditis slight clubbing petechiae and retinal haemor rhages The earlier a case is recognized the better the outlook and the diagnosis of an infected ductus should be at any age an indication for urgent operation because progressive exhaustion and pulmonary infarcts increase the surgical hazards and make the operation technically more difficult

Electrocardiogram As might be expected with a lesion that is extracardiac the electrocardiogram is normal in appearance There may be a tendency to left axis if the shunt is at all large and in older cases there may be a frank left axis A right axis is most uncommon and should lead to a revision of the diagnosis although Steinberg *et al* (1943) found right axis deviation in a number of their cases

Radiology The radiological picture of the patent ductus is in fluenced by the presence or absence of other anomalies of the heart Even when the patent ductus is present as a sole abnormality it by no means follows that the radiological picture is characteristic The radiograph is influenced by the age of the patient and by the nature of the ductus itself and particularly by the size of the orifice of the ductus and the shunt that it permits The heart is only slightly enlarged and this may be evident by an increased curvature of the left ventricular

arc To find a large heart is rare and the possibility of infective endo carditis or the association of other abnormalities is suggested by its presence The right auricle is frequently enlarged Where there is a large shunt there is considerable dilatation of the pulmonary artery the dilatation extending to its branches (fig 45) This pulmonary artery dilatation may in some cases assume aneurysmal proportions On the screen there is vigorous pulsation of the aorta and pulmonary artery the latter showing a hilar dance or Assmann s (1924) sign



FIG 45 Patent ductus arteriosus in a girl aged 25 The pulmonary artery is greatly dilated The lesion is without symptoms other than those of a neurosis

The typical silhouette shows a normal sized or slightly enlarged heart with normal aorta There is an increased fullness of the pul monary arc which in well marked cases may be a prominent con vexity on the left cardiac border This prominence has been named the X ray cap of Zinn (1898) and was one of the earliest features recorded in the radiography of the heart As indicated above cases with marked thrill and murmur and in every way clinically typical do *not always give a characteristic X ray picture* Pulmonary congestion is common The value of a dilated pulmonary arc in the presence of typical signs is at once evident for it affords confirmatory evidence There are other congenital conditions where notable dilatation of the pulmonary arc is present such as pulmonary stenosis with a closed septum and interauricular septal defects The recognition of these on clinical grounds should not cause great difficulty

Other lesser radiological signs have been described The left pul monary artery may appear to be larger than the right owing to the direction of flow from the ductus Roesler (1937) mentions a fleck of

calcium in the aorta at the site of attachment of the ductus Steinberg Grishman and Sussman (1943) with angiocardiography found a constant dilatation of the descending aorta just distal to the isthmus and best seen in the left anterior oblique position They confirm the presence of varying degrees of left ventricular dilatation Donovan Neuhauser and Sosman (1943) and Eppinger Burwell and Gross (1941) mention dilatation of the left auricle but this is not always confirmed by angiocardiography although it undoubtedly does occur Chavez Dorbecker and Celis (1947) by their method of direct intracardiac angiocardiography have shown by several films at intervals of two seconds back flow filling of the pulmonary artery at the time that the aorta fills a circumstance only possible if there is a fistula between the two vessels This may well be a decisive point in a doubtful case

COURSE AND PROGNOSIS All will agree that from the point of view of childhood the prognosis is good and the condition appears to be perfectly compatible with a normal school life the amount of exercise permitted being often the same as in normal children unless there are evident signs of distress Serious heart disability may appear in the third decade (Bullock Jones and Dolley 1940) In Abbott s (1937) series one fifth of the patients died in the neonatal period and the average age at death of the remainder was 24 years These figures are perhaps unnecessarily gloomy and mainly refer to cases with symptoms or infection that have died in hospital Bullock Jones and Dolley collected thirty cases who were more than three years old and therefore of interest to the surgeon They found that 50 per cent died before the age of 30 and 71 per cent before 40 In 87 per cent the cause of death was cardiac either infection or failure Shapiro and Keys (1943) found the average age at death to be 30 in the male and 35 in the female 40 per cent dying of bacterial endocarditis and 30 per cent of con gestive failure On the other hand Wilson and Lubschez (1942) observed thirty two cases for a period of twenty years and saw no deaths from infection or failure

If all these statistics are taken into consideration the general con clusion in our present state of knowledge must be that the prognosis is rather poor and that survival to middle life is accompanied by considerable cardiac incapacity

None the less it is doubtful if the last word has been said in the matter of prognosis Our ideas are mainly based on post mortem cases and take little account in general of the living case. As mentioned above the adult case of patent ductus is a rare visitor to the clinic and the condition appears to be equally rare in the routine post mortem examination It therefore seems that if post mortem room technique is impeccable there must be spontaneous closure of the ductus more often than is generally realized or cases are missed when failure super venes and the characteristic physical signs disappear

Surgical Treatment of the *Patent Ductus Arteriosus*

Holman (1940) showed that in a peripheral arteriovenous fistula surgical correction of the leak resulted in a rise in diastolic pressure and a decrease in the size of the heart From this there is reason to infer that surgical treatment of the patent ductus arteriosus might improve the circulation check the tendency to underdevelopment and prevent the occurrence of congestive failure at an early age Similarly the risks of infective endocarditis might be minimized and degenerative changes in the pulmonary artery wall where the stream from the ductus impinges would be prevented

Munro (1907) was the first to suggest ligature of the ductus In 1938 the operation of Graybiel and Strieder failed because of technical reasons Gross and Hubbard (1939) reported the first successful ligature and Bourne and Tubbs (1941) first successful ligature and cure of an infected case in 1939 Shapiro and Keys (1943) were able to collect 140 operated cases of whom thirty three were suffering with bacterial endocarditis Of the 107 non infected cases operation was successful in eighty one and nine died at or soon after operation In the thirty three infected cases the operation was successful in twenty and five died at operation Since then with improvements in technique and experience the mortality is lower and Crafoord (1944) places the operative death rate at about 5 per cent Shapiro and Johnson (1947) analysing 525 uninfected cases in which operation had been performed by forty six surgeons found a mortality rate of 4 9 per cent Re canalization occurred in 8 7 per cent and because of this suggest that ligation of the duct is obsolete and should only be performed where section is impossible

In the management of these cases each individual case should be considered on its own merits Because of the rather striking liability to infective endocarditis focal sepsis should be carefully eradicated Particular care should be devoted to the teeth and tonsils Dental extractions in the presence of infected gums and tonsillectomy should be preceded and followed by full doses of penicillin

Errors in diagnosis will be less frequent if only those cases with a typical machinery murmur are submitted to operation The justification of operation resides in the fact that about 50 per cent of the cases die before the age of 30 Operation with its comparatively small mortality rate offers a chance of cure with the prevention of infective endocarditis The chances of recurrence are remote if the operation has been sufficiently radical and recanalization of the ductus rarely occurs in those cases where there has been a simple ligature Apart from this from a social point of view the case of patent ductus is liable to rejection from a public service such as the police or armed forces and cannot pass examinations for life insurance or superannuation

Operation should be contemplated between the ages of five and ten

for this is the time when the largest number of cases are recognized at school entrance examinations At this age it is possible to use an endotracheal tube for the necessary anaesthesia There is also less likelihood of gross sclerotic changes in the ductus and further the ductus is likely to be longer than at a later age The experience of surgeons suggests that up to the age of 15 the operation is well tolerated and a number of cases have been operated upon at later ages

In regard to the infected case the presence of bacterial endocarditis should be an indication for operation at any age and offers the best chance to the patient Operation should not be delayed and should be preceded by and accompanied by intensive treatment with penicillin

The general indications for operation are retardation of growth and development bacterial endocarditis cardiac insufficiency and failure (Gross 1939 Burch 1944 Tubbs 1945) Recovered bacterial endocarditis is also an indication because these cases have some tendency to relapse

RESULTS OF OPERATION The immediate effect is a rise of the diastolic pressure to a physiological level The pulse pressure becomes normal and the vigorous pulsation in the neck disappears The characteristic murmur is abolished The size of the heart diminishes This is more noticeable in young subjects where dilatation rather than hypertrophy has been the predominant factor in the existing cardiac enlargement Where there have been actual hypertrophic changes the heart may alter little in size but with growth of the individual a more normal size will be evident in a year or two Eppinger Burwell and Gross (1941) have shown that a fall of at least 50 per cent of the left ventricular output occurs after ligature

In regard to the infected case Touroff (1942) showed that even without penicillin ligature resulted in cure in a high proportion of cases and that a negative blood culture might be obtained immediately after operation Presumably these cases are those in which infection remains limited to the ductus itself and to the pulmonary artery The addition of arterial blood to the pulmonary blood creates a milieu rich in oxygen in which organisms and vegetations flourish The impinging stream from the ductus breaks off particles which become emboli in the lungs These events cease when the ductus is ligatured and the patient tends to recover rapidly With the aid of penicillin cure is almost certain It is only when infection involves the mitral or aortic valves (shown by systemic emboli) that results are not good by purely surgical means However with penicillin the prognosis in this group of cases is as good as in any other case of infective endocarditis

Lastly many patients remark the absence of dyspnoea which had been habitual to them and passed unnoticed until after operation

CHAPTER X

DEFECTS OF THE AURICULAR SEPTUM

HISTORICAL NOTE The earliest mention of the foramen ovale is attributed to Galen who was aware that the foramen ovale possessed a valve and closed at birth and that in the foetus the caval blood passed through the foramen ovale into the left auricle These observations lapsed into oblivion and in 1557 the foramen ovale was rediscovered and described by Aranzio Botallo (1564) again described the foramen and renamed it the ductus Botallo without any justification whatsoever He did not describe the ductus arteriosus as is so often *erroneously claimed The first illustrations of the foramen ovale are* those of Laurens (1599) unfortunately reversed but adequate to show *that the structure was known Harvey (1628) mentions the passage of* blood through the foramen into the left auricle of the foetus Gassendi (1640) noted the existence of vestiges of the foramen ovale in the adult at a time when the question of the perviousness of the septum of the heart was vitally necessary to the concept of the circulation of the blood Richard Lower (1699) described two vena cavae and seems to have recognized that the inferior caval stream diverted by the tubercule that bears his name passed mainly through the foramen ovale of the foetus into the left auricle Senac (1745) recognized the foramen ovale in adult life stating that it was three times as common in the female as in the male Burns (1813) held along with the naturalist Buffon that the subject of a patent foramen ovale could stand immersion in water longer than others and a diver to be efficient at his work should *possess this anatomical adjunct* This ingenious view presumably arose from the concept of the foetus bathed in its liquor amnii Meckel (1812) dismissed a patent foramen ovale as clinically negligible Peacock (1866) observed that defects might be anywhere in the septum and might even be so situated as to allow all four cavities of the heart to intercommunicate He also described a case with coincident mitral stenosis Rokitansky (1875) presented the first complete description of the types of auricular septal defect and initiated modern embryological research with its concept of primary and secondary septa The subsequent work of a host of other anatomists and pathologists *is complementary to his* general pattern of an auricular septum formed by the union of incomplete septa

On the clinical side mention may be made of the fundamental work of Abbott (1927 1932) Lutembacher (1916 1925) McGinn and White

(1933) Rosler (1934) Taussig (1938) and Bedford Papp and Parkinson (1941) all of whom have made outstanding contributions to a clinical picture that has now become established

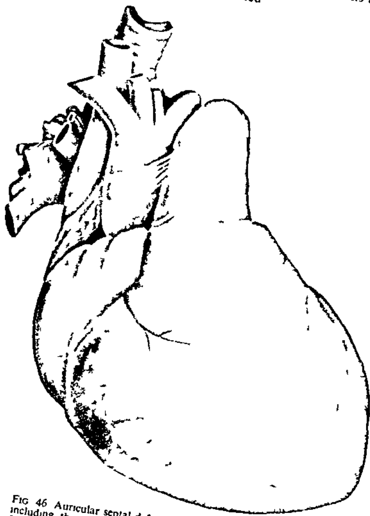


FIG 46 Auricular septal defect Anterior surface of the heart including the right apex is formed by the right chambers Pulmonary artery greatly enlarged (Bedford Papp and Parkinson)

EMBRYOLOGY The stages in the development of the auricular septum may be briefly recapitulated as they are important in relation to the genesis of the various defects enumerated below (fig 46) The first

indication of septation is the appearance of the septum primum which grows from the roof of the common atrium at about *the fourth week* A large gap the foramen (ostium) primum is present just above the auriculoventricular orifice and interventricular septum and beneath the free margin of the septum primum The ostium primum is gradually obliterated while coincidently a gap appears in the upper part of the septum primum the ostium secundum To the right of the septum primum another septum grows down This is the septum secundum and its free margin forms the annulus ovalis The disposition of the two septa is such that there exists a potential communication between the two auricles the foramen ovale which may have the status of a valve like slit or may assume more important proportions The free edge of the septum primum persists as the valve of the foramen ovale

It is therefore obvious that there are a number of possible congenital abnormalities These are an absent auricular septum persistent ostium primum and persistent ostium secundum (often erroneously referred to as a widely patent foramen ovale) Rarely there may be a premature closing of the foramen ovale A persistent ostium primum with abnormality of the endocardial cushions of the atrial canal may result in an ostium atrioventriculare commune Defects of the auricular septum may therefore be readily explained on an embryological basis particularly if Patten s (1938) dictum is borne in mind that congenital defects may result alike from overgrowth or developmental arrest

Apart from these purely embryological views as to the genesis of the auricular septal defect itself the French school consider the defect to be a small part of a more complex congenital lesion comprising large right cavities and a dilated pulmonary artery together with a hypoplasic aorta and left ventricle (grosse pulmonaire petite aorte) Such a defect is considered to be latent and only important when complicated by other lesions such as mitral stenosis or hypertension which by *causing altered pressure relationships* produce secondary changes leading to failure (Laubry and Lenegre 1939 de Balsac 1939) An unequal division of the bulbus cordis does occur (Assmann 1928 Oppenheimer 1933) and may produce a clinical and radiological picture practically identical with that of the auricular septal defect There are objections to the French view for in the auricular septal defect autopsy may show the aorta to be of normal size with a large *pulmonary artery* having hypoplasic walls (Brenner 1935) In those *cases where there* is hypoplasia of the aorta the small calibre of the aorta may be ascribed to a diminished blood volume reaching *the left* ventricle as has been described in mitral stenosis developing in early life

FUNCTION OF THE FORAMEN OVALE The function of the foramen ovale has been illuminated by the researches of Barclay and his collaborators (1944) who used injection of radio opaque *substances and*

cinematographic methods They have shown without a doubt that in the foetus four fifths of the inferior caval blood stream is diverted through the foramen ovale the stream bifurcating upon the limbus of the fossa ovalis or as they have named it the crista dividens whence it passes into the systemic circulation of the foetus Only the superior caval stream is directed into the pulmonary circuit for in foetal life the pulmonary circulation obviously does not require that amount of blood necessary for its respiratory function in post natal life

It should not be assumed that closure of the foramen ovale is a sudden mechanical event occurring at birth with a startling change in the dynamics of the heart Scammon and Norris (1925) have shown from autopsy records that but 1 per cent are completely closed in the first week 53 per cent are closed at the end of a year At twenty years or over 28 per cent still remain patent For this reason slit like patency should be considered as a normal anatomical variant rather than as being of any direct clinical significance The possible clinical implications of slit patency are discussed below

TYPES OF DEFECT OF THE AURICULAR SEPTUM

There are several types of interauricular septal defects depending upon their position and embryological origin The more important types are

(1) *Absent or Rudimentary Septum* (cor triloculare biventriculare) This anomaly results from an arrest in the early stages of the development of the septum (see page 227)

(2) *Persistent Ostium Primum* This anomaly is due to failure of union between the septum primum and the fused endocardial cushions of the atrial canal As a result there is a large gap at the base of the inter auricular septum (fig 47) and it will be appreciated that an extreme degree of persistence of the ostium primum merges into the cor tri loculare biventriculare Almost invariably associated is a cleft anterior cusp of the mitral valve The anterior cusp of the mitral valve is divided into two halves each half runs upwards from its papillary muscle and converge on the lower margin of the defect at which site the anterior half overlaps the posterior A similar anomaly may some times involve the tricuspid valve as in Bedford Papp and Parkinson s case (1941) Additionally the foramen ovale may be open or closed and may be recognized above the defect at the base of the septum Of added interest is the frequency noted by Abbott (1927) with which a persistent ostium primum is associated with mongolism Occasionally heart block may be associated as in the case of Wallgren and Winblad (1937)

(3) *Persistent Ostium Secundum* The defect is the result of failure of development of the septum secundum so that a gap exists in the upper and posterior part of the septum above the fossa ovalis The anomaly is frequently combined with abnormalities of the pulmonary veins and

of the inferior vena cava (Rokitansky 1875 Duff 1938 Ash 1939) Thus in several reported cases the right pulmonary veins have opened *into the right auricle* or into the superior vena cava and the orifice of the inferior vena cava may look into both auricles In other cases the superior vena cava has lain astride the defect or a left superior vena cava has been present as in the case of Chase (1938) This anomalous condition of the veins has been supposed by some to be the cause of persistence of the ostium secundum because of the extra amount of



FIG 47 Persistent foramen primum The mitral and tricuspid valves are united at the upper margin of the interventricular septum (Specimen in the R C S Museum)

blood received by the right auricle This hypothesis does not receive entire confirmation for in other cases there have been no anomalies of the veins as in the case of Abbot and Kaufman (1910) In this latter case it seems that there was a true failure in development of the septum secundum The identification of this defect should present few difficulties because owing to failure of the septum secundum there is no restriction of the foramen ovale and the septum primum in the later stages of their development Consequently the valve membrane and the limbus of the fossa ovalis are absent in a true defect of the category The term widely patent foramen ovale should be applied only to a distention of a functionally patent foramen ovale without congenital septal defect

(4) *Patent Foramen Ovale* Valvular or slit like patency of the foramen ovale is a common finding in the post mortem room and is without clinical significance It occurs in at least 20-30 per cent of cases and it should be recognized as an anatomical variant of interest to the anatomist but clinically unimportant Inspection of the anatomical specimen shows that the orifice or potential orifice does not gape but

is kept closed by the overlap of the valvula foraminis ovalis and the annulus ovalis in the left and right auricles respectively Such slit like patency is quite without signs or symptoms and is in general without significance There is however evidence to show that in the event of an increase in pressure in the right auricle occasioned by a pulmonary embolus hypertensive heart failure or mitral stenosis the potential patency may become opened up and actual so that cyanosis may develop or a paradoxical embolus be allowed to pass (fig 48) This

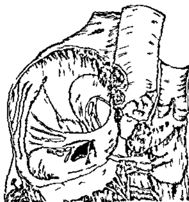


FIG 48 Patent foramen ovale Female aged 20

latter is a very rare event but one which may be reasonably inferred or suspected from time to time Slit like patency should not be confused with failure of development of the septum secundum which is characterized by absence of the annulus and the valve membrane The foramen ovale may also remain patent owing to conditions favouring a rise in pressure in the right auricle after birth as in pulmonary stenosis Similarly tricuspid atresia entrains a patent foramen ovale of considerable dimensions if a circulation is to be maintained A few cases have been recorded where there were multiple defects of the septum especially of the valvula

Although it is obvious that there is usually an embryological explanation of most auricular septal defects it is to be regretted that many post mortem reports are confusing and inexplicit as to the specific anatomical lesion to which they refer A defect of 1 cm diameter is usually considered to be significant and the starting point of pathology

It must be emphasized that although the site of the defect may occasionally determine certain peculiarities in the clinical picture the symptoms and physical signs depend as a whole upon the size of the defect and not upon its site

INCIDENCE An auricular septal defect is the commonest of all

congenital heart abnormalities As a combined lesion its incidence may be as high as 85 per cent of all cases of congenital heart disease It is impossible to enumerate all the defects with which it may be associated but in many abnormalities the defect serves an integral part in maintaining a circulation as in the valvular atresias and more complex anomalies Its incidence as a sole lesion is given as 7 25 per cent by Bedford Papp and Parkinson (1941) the latter figure perhaps being unduly high It must be remembered that incidence at autopsy may not necessarily correspond with clinical incidence but there is reason to believe now that the clinical picture permits an almost certain diagnosis that these figures will undergo alteration The defect as a sole abnormality or in association with mitral stenosis appears to be much more common in females but Burrett and White (1945) believe that both sexes are equally affected

PATHOLOGY The most striking changes in the auricular septal defect are in relation to the right side of the heart (fig 46) There is hypertrophy and dilatation of the right auricle and ventricle and dilatation of the pulmonary artery and its branches Hypoplasia of the aorta is frequently present and this was considered by Rokitansky to be the primary abnormality which by raising pressure in the left side of the heart would tend to keep the foramen ovale open Against this theory is the frequency with which a congenital aortic stenosis is unaccompanied by a defect of the septum Further it is possible for the aorta to appear small in comparison with the pulmonary artery yet to be of normal calibre

As a rule the heart as a whole appears to be greatly enlarged as in Roesler s (1934) girl of 14 where it weighed 800 grams Roesler s analysis confirms this general enlargement in the sixty two cases of his series and suggests that the larger the defect the larger the heart is likely to be The heart has a rather square appearance the right cavities forming the anterior surface and apex of the heart There is a marked disparity between the calibres of the aorta and pulmonary artery and Rosler estimates their respective ratios as 2 3 The pulmonary artery is dilated the dilatation extending to its smaller branches (fig 51) Atheroma of varying degree may be present and likewise recent or old thrombosis Associated with these changes there may be pulmonary infarction There may also be evidence of paradoxical embolism with a ribbon like clot extending through the defect This is a very rare occurrence The visceral changes are those of chronic congestion with an enlarged liver Ascites and peripheral oedema may be present pleural effusion is rare and is most likely in the case complicated by mitral stenosis The co existence of mitral stenosis does not greatly alter the above general appearance save that the heart tends to be even larger and the disproportion between right and left chambers more exaggerated The left auricle is small the septal defect tending to prevent its dilatation

SECONDARY PATHOLOGICAL EFFECTS The constancy of right sided dilatation and hypertrophy is impressive and is in marked contrast to the smallness of the left side of the heart and aorta These changes are the natural consequence of a shunt which passes from left to right through the defect increasing the load on the right side and diminishing that on the left The additional burden of a mitral stenosis does no more than to emphasize those changes already present The left auricle is of particular interest for contrary to pathological experience of mitral stenosis with an intact septum where the left auricle is hyper trophied and dilated it remains small and relatively undilated when there is a septal defect

It has always been assumed that hypertrophy and dilatation of a chamber of the heart are an expression of an increased load or work done by that chamber Equally it has been assumed that pressure differences between the left and right auricles are the cause of a shunt from left to right yet anatomically in these cases there is no hyper trophy of the left auricular wall This suggests that one effect of a septal defect of significance is to equalize pressure in the two auricles This point being conceded it is necessary to find some explanation other than a shunt caused by pressure differences to account for the constant right sided hypertrophy The recent work of Uhley (1942) has shown that the plane of the auricular septum is not vertical as is commonly supposed but nearly horizontal the left auricle being on a slightly higher plane than the right with the septum forming its floor Because of this there is a constant gravitational flow through the defect and only when the subject is recumbent and the septum vertical is there any real reduction in this flow A mitral stenosis obstructing the egress of blood into the left ventricle will emphasize this flow and tend to cause more marked changes in the right side It seems therefore likely that the first effect of an interauricular septal defect is to equalize intra auricular pressures Subsequently gravity alone will lead to a gradual enlargement of the right side of the heart This process may be hastened by the intervention of mitral stenosis Uhley s experiments are convincing in this respect and accord well with the observed clinical facts of a long latent period before symptoms actually develop and their emphasis when mitral stenosis occurs This theory has not received confirmation in experiments with a catheter in the right heart carried out with the subject inverted by Brannon Weens and Warren (1945) These authors found the right ventricular pressure in two un complicated cases was 40 and 41 mm Hg respectively the output of the right ventricle was twice that of the left and the right atrial blood had a higher oxygen content than blood from the superior and inferior vena cava This latter observation proves the existence of a shunt

ASSOCIATION OF ACQUIRED VALVULAR LESIONS The auricular septal defect is by far the commonest form of congenital heart disease with

which acquired valvular disease of the rheumatic type may be associated Roesler (1934) found that three quarters of his collected cases had evidence of rheumatic valvular disease The mitral valve is most commonly involved alone although coincident changes have been reported in the aortic or tricuspid valves

Mitral Stenosis (Lutembacher's Disease)

Mitral stenosis is the commonest valvular lesion associated with an auricular septal defect The first observation has been credited to Louis (1826) but a case was described by Corvisart (1814) Other early examples are those of Mayne (1884) and Peacock (1860) It was present in eleven of the twenty cases of *Rokitansky (1875) Griffiths (1906)* case was exceptional because of the presence of infective endocarditis Abbott (1915) and Lutembacher (1916) stressed the frequent coincidence so that it became known as Lutembacher s Disease McGinn and White (1933) collected twenty three cases from the literature and added one pointing out the overwhelming preponderance of females Lutembacher (1936) revived interest in the condition and cases were published by Cossio and Berconsky (1936) Cossio and Arana (1937) Battro and de la Serna (1937) Taussig *et al* (1938) to cite a few examples Recently Bedford Papp and Parkinson (1941) reported four autopsied and four clinical cases in a group of fifty three patients with an auricular septal defect Burrett and White (1945) who have reviewed the thirty one autopsied cases since Roesler s series have come to the conclusion that when Roesler s and their own series are combined no less than 53 8 per cent of cases of auricular septal defect have mitral stenosis at autopsy For this reason a coexisting mitral stenosis should be suspected in every case of the auricular septal defect despite the absence of any definite clinical indication of its presence This is well shown by the experience of Bedford Papp and Parkinson who found mitral stenosis in four of ten autopsies and clinically in only four of their remaining forty three cases

Opinion varies as to the origin of the mitral stenosis Some have favoured a congenital origin postulating a foetal endocarditis of the mitral valve Yet others notably in France have thought the mitral stenosis to be part of a more complex congenital abnormality involving the left side of the heart so that the aorta is small and the pulmonary artery large Narrowing of the mitral valve from any cause would tend to prevent closure of the foramen ovale The documented cases of congenital mitral stenosis are conspicuous by having their foramen ovale closed or very small All the available evidence points to an acquired valvular lesion But if acquired valvular disease is common as we think it is there is yet no positive evidence to confirm that its incidence is any higher in this group than it is amongst the general population at large None the less its incidence in this particular and

restricted field of congenital heart disease is very much higher than Gelfman and Levine s (1942) estimated incidence of acquired rheumatic valvular disease in 13 per cent of congenital heart cases as a whole This requires some explanation and a very accurate delineation of the type of associated auricular septal defect A clinical history of rheumatic fever or chorea is rare in the auricular septal defect An absence of rheumatic antecedents should not necessarily be disconcerting in view of the large number of cases of mitral stenosis ordinarily encountered in any group of young pregnant women A personal view was quite simply expressed in 1943 (Bradshaw Lecture R C P) by stating that were it not for the mitral stenosis little would be heard of the auricular septal defect The extraordinary coincidence of mitral valvular disease with this defect as shown by post mortem incidence forces the view that mitral stenosis is most often the primary event which renders a potentially patent foramen ovale into a defect of significance

Aortic valvular disease was present in the cases of Jacobius and Moore (1938) and Taussig Harvey and Follis (1938) Pericarditis was mentioned by Rokitansky (1875) Battro and de la Serna (1937) Cossio and Arana (1937) Bedford Papp and Parkinson (1941) and others

Bacterial endocarditis is almost unknown despite the presence of acquired valvular disease or other congenital defects Four cases are known to have had associated bacterial endocarditis and are those of Griffith (1906) Goetsch (1938) Jacobius and Moore (1938) and Bedford Papp and Parkinson (1941) To these may be added one in a dog reported by Osler Abbott (1941) In congenital heart disease as a whole 15 5 per cent of cases develop bacterial endocarditis In a series of 453 autopsied cases of congenital heart disease collected by Gelfman and Levine (1942) 6 5 per cent had infective endocarditis There were 179 cases with a significant auricular septal defect as a primary or complicating abnormality and all were exempt from infection It is interesting to note the type of defect in this series A persistent ostium secundum in 140 cases persistent ostium primum twenty multiple defects nine and absent auricular septum in ten In 181 cases above the age of two the incidence of bacterial endocarditis was 16 6 per cent There were forty five cases with a significant auricular septal defect in this group all exempt from bacterial endocarditis It is obvious that sites of stress and strain or of vigorous shunts have no counterpart in the auricular septal defect where the shunt is mainly passive and gravitational Even when mitral stenosis is present left intra auricular pressure is not notably raised because of the pressure equalizing effect of deflection of the blood through a wide septal defect

CLINICAL PICTURE Abbott (1934) has stated that the clinical picture is dominated by the presence of hypoplasia of the aorta Depletion of the systemic circulation by the combined effects of a persistent shunt

from left to right and underfilling of the left side of the heart leads to pallor of the skin a rather slender build and delayed signs of puberty Dwarfism and infantilism have been reported in this condition The association of mongolism with a persistent ostium primum has been mentioned elsewhere It appears as more cases are reported that these possible physiological effects and associated abnormalities have been over emphasized

The abnormality may be entirely latent until the third decade or later when symptoms often appear for the first time and are those of breathlessness and failure Dyspnoea is generally present and may be so habitual to the patient that it is not complained of It is often only slight and despite an enlarged heart there may be a reasonable capacity for exertion Cyanosis is inconstant and may appear first with exertion or in imminent or actual failure The cyanosis is of the type of cyanose tardive (Bard and Curtillet 1899) and is consequent upon reversal of the shunt which then flows from right to left through the defect Cyanosis may also appear during intercurrent infections such as pneumonia or be associated with haemoptysis when thrombosis occurs in pulmonary branches Established cyanosis in the absence of failure is very rare and should suggest the presence of some complicating abnormality Clubbing is likewise inconstant and is proportional to the degree of and duration of cyanosis Haemoptysis may also be due to an accompanying mitral stenosis Hoarseness due to pressure of the enlarged pulmonary artery on the laryngeal nerve has been described by Wahl and Gard (1931) Erlanger and Levine (1943) and Burrett and White (1945)

As regards the purely cardiac signs a precordial bulge may be present but is only a sign of cardiac enlargement occurring at an early age The apex beat is forcible and displaced to the left and in the absence of aortic incompetence or hypertension is an important sign (Rosler 1934) There are no characteristic murmurs and a murmur may be entirely absent Commonly there is a systolic murmur at the apex or in the pulmonary area The murmur is not produced in the defect itself and according to Cossio *et al* (1938) is probably produced at the less distensible pulmonary ring which intervenes between a dilated pulmonary artery and conus The pulmonary second sound is emphatic and there is often a palpable diastolic shock in the pulmonary area A systolic thrill may be palpable at the same time Ribbon dullness similar to Gerhardt s dullness in the patent ductus may also be present The combination of cardiac enlargement to the anterior or mid axillary lines with signs of pulmonary artery dilatation is very suggestive In other cases a systolic bruit may be due to a dilated tricuspid orifice The characteristic bruit of mitral stenosis may be present at times but in most cases it is usually not heard and mitral stenosis thus remains silent owing to the low intra auricular pressure

and shunt caused by the defect In a proportion of cases a diastolic murmur of pulmonary incompetence may be heard in the pulmonary area and along the left sternal border The blood pressure is generally low unless the patient is within the age group where hypertension occurs

Auricular fibrillation may be present and its occurrence is of interest owing to its extreme rarity in other forms of congenital heart disease It is due to the complicating presence of mitral stenosis or incompe

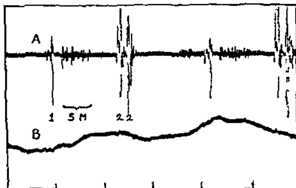


FIG 49 Atrial septal defect Patient aged 9 with typical fluoroscopic appearances A logarithmic phonocardiogram recorded from the second intercostal space at the left sternal edge B apex cardiagram 1 First sound S M systolic murmur 2 2 split second heart sound gap 0 05 sec Time intervals 0 ? sec (Dr M Matthew s case)

tence and no case of fibrillation has so far been reported in the pure auricular septal defect unless there has been hypertension or other added pathology Flutter is rare but cases have been reported by Amberg and Willus (1926) and by Bedford *et al* (1941)

At the time of writing little is known of the exact clinical picture of the auricular septal defect in childhood and most cases are unrecognized There is reason to believe that the development of cardiac catheterization and angiocardiography will remove many uncertainties in diagnosis Sometimes an X ray of the chest taken during a mass survey or for some intercurrent infection may evoke the first suspicion of cardiac abnormality A systolic murmur may or may not be present In the author s experience if a murmur is present it is in no way characteristic and may be quite indistinguishable from the systolic murmur of other types of acyanotic congenital heart disease Indeed its audibility in the area of auscultatory romance may well lead to its dismissal as of no account Not infrequently the pulmonary second sound is abnormally loud and reduplicated (fig 49) and this may lead to further investigation and the radiological display of a prominent

pulmonary conus and a large right branch The point is that the diagnosis is elusive in childhood and it may be years before there is a characteristic clinical picture Auricular flutter paroxysmal tachycardia acrocyanosis in the very young and failure of normal development are possible findings which may raise suspicion of the defect

Radiology The principal radiological sign is enlargement of the pulmonary artery and its branches (Assmann 1928) In the anterior view the heart is always enlarged to the left and often to the right



Fig 50 Defect of auricular septum in woman aged 35 Radiograph showing much enlarged heart and aneurysmal pulmonary artery its right branch formed a pulsating mass in right lung root

Battro and De la Serna (1937) and Cossio and Arana (1937) considered enlargement to the right to be an important radiological sign Bedford Papp and Parkinson (1941) suggest that the right auricle tends to bulge forward and to the left displacing the enlarged right ventricle The enlarged right ventricle forms the rounded left border of the heart A prominent bulge of the pulmonary arc is the most striking finding and may assume aneurysmal proportions (fig 50) This is largely due to dilatation of the trunk of the pulmonary artery although the left branch may also be a component of and emphasize the arc The pulmonary branches are always enlarged and the right branch which is not covered by any part of the cardiac silhouette is dense and comma shaped On the screen pulsation of the pulmonary artery and its branches is often prominent, the so called hilar dance (Pezzi 1925 1932) Absence of pulsation suggests the presence of pulmonary artery thrombosis Such pulsation has been described as being due to pulmonary incompetence but that is not always so The aortic knob is small or absent in most cases and when mitral stenosis is present there is likely to be right

auricular dilatation In the oblique view the left auricle is not enlarged

In a few cases (Roesler 1934 Taussig *et al* 1938 and Joly 1939) the heart is pyriform in shape and suggests a pericardial effusion (Bedford *et al* 1941) Angiocardiography is of little value in the uncomplicated defect

ELECTROCARDIOGRAM Right axis deviation either of itself or as bundle branch block of the old or newer types is almost always present Large P waves in one or more leads are common and a bifid P wave

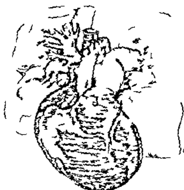


FIG 51 Same case pulmonary artery and main branches dilated right ventricle much enlarged and aorta hypoplastic

may be present in the absence of mitral stenosis The PR interval may be increased (Bedford *et al* 1941) and there is evidence to show that in some cases the PR interval undergoes progressive increase with age All degrees of widening or splintering of the QRS complex are common ranging from simple prolonged intraventricular conduction down to right bundle branch block of the commoner or newer types (fig 52) Incomplete bundle branch block is commoner than the complete form A widened QRS suggesting right bundle branch block has been described by Bedford and Brown (1937) Routier and de Balsac (1938) and was thought to be of diagnostic importance by Routier Brumlik and Malinsky (1940) These latter suggest that this change is not a congenital abnormality *per se* but is induced by the progressive myocardial changes accompanying the gradual hypertrophy of the heart during the course of the disease The T wave in lead 2 or 3 or both is frequently inverted when a widened QRS is present but T wave inversion may be present with normal ventricular complexes T wave inversion is said to be an expression of right ventricular strain (Barnes and Whitten 1929 Brumlik 1937) Complete heart block has been occasionally recorded in persistent ostium primum (Wallgren and Winblad 1937)

Auricular fibrillation occurs with some frequency and the

interauricular septal defect is the only congenital lesion where it is likely to occur It is thought to result either from the associated rheumatic mitral stenosis or from increasing age of the patient rather than from the actual congenital lesion itself In all the reported cases it has been associated with mitral stenosis in the younger age groups and there is no record of fibrillation in the *pure uncomplicated defect below the* age of 50

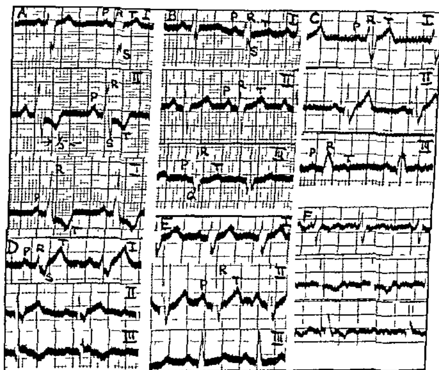


FIG 52 Electrocardiograms from six cases of auricular septal defect It will be seen that conduction defects are frequent and that various types of typical and atypical bundle branch block may be present In case F there was auricular fibrillation

DIAGNOSIS The diagnosis of an *interauricular septal defect is mainly* radiological The gross dilatation of the pulmonary artery and its right branch together with enlargement of the heart is characteristic From a purely clinical point of view *enlargement of the heart in the absence* of aortic incompetence or hypertension is very suggestive In addition there are the signs of dilatation of the pulmonary artery visible and palpable pulsation *in the left inter spaces* a diastolic shock and per haps a systolic thrill A pulmonary diastolic murmur may be present Cyanosis is absent unless failure is present or imminent and although

dyspnoea is present it seems much less than would be anticipated from the clinical and radiological signs The electrocardiogram shows a right axis possibly with negative T waves in leads 2 and 3

Differential diagnosis is from those congenital or acquired conditions in which the pulmonary artery is enlarged It must never be forgotten that an auricular septal defect is often combined with other congenital lesions

Mitral stenosis may cause difficulty if the pulmonary artery is enlarged or if there is relative pulmonary incompetence However enlargement of the left auricle and displacement of the barium filled oesophagus will exclude an auricular septal defect In addition mitral stenosis will more often present a straight left border of the cardiac silhouette Likewise there is apt to be a general pulmonary congestion conspicuously absent in the auricular septal defect Finally the characteristic late diastolic murmur of mitral stenosis is present As mentioned above it is wise to suspect mitral stenosis in every case of interauricular septal defect The presence of auricular fibrillation in a case which presents all the signs of the septal defect both clinical and radiological and below the age of 50 is almost certain to have mitral stenosis present

A patent ductus arteriosus rarely causes difficulty The heart is not greatly enlarged a machinery bruit is characteristic and the electrocardiogram shows a normal or left axis deviation Only in those cases where the patent ductus is complicated by pulmonary artery sclerosis and pulmonary hypertension is there any real difficulty In pulmonary stenosis there is a characteristic harsh murmur and systolic thrill in the pulmonary area The pulmonary branches are not enlarged although the trunk may be dilated The aortic knuckle is readily visible and there is not extreme cardiac enlargement

The maladie de Roger is unaccompanied by gross cardiac enlargement or by a dilated pulmonary artery There may be a straight left border to the heart The electrocardiogram shows a normal axis in most cases

Pulmonary artery dilatation secondary to chronic lung disease shows the signs of the causal condition Clubbing of the fingers may be prominent

Primary pulmonary artery disease may be most difficult but suggestive points are cyanosis of long standing clubbing and a positive Wasserman reaction

Primary dilatation of the pulmonary artery due to unequal division of the common trunk may present the greatest difficulty of all as the clinical and X ray picture may be identical with the true auricular septal defect Exact recognition may depend upon post mortem examination Primary dilatation is extremely rare and probability in a doubtful case would favour the interauricular septal defect (Bedford *et al*) Brandon Weens and Warren (1945) and others have shown by

heart catheterization that in the auricular septal defect the oxygen content of blood in the right auricle is appreciably higher than in blood from the superior or inferior vena cava This affords a possible means of distinction between septal defect and primary dilatation of the pulmonary artery

COURSE AND PROGNOSIS Symptoms may be slight or absent until adult life and many patients appear to be capable of normal physical activities Others are delicate and lead restricted lives

Cyanosis and dyspnoea gradually develop and ultimately the case presents the general picture of right heart failure Similar symptoms may appear suddenly during some intercurrent disease Death is usually the result of cardiac failure often with auricular fibrillation A few cases may die of pulmonary tuberculosis but infective endocarditis is of extreme rarity and a pathological curiosity Paradoxical embolism (Chapter I) is a classical manifestation but occurs rarely as in the cases of Thomson and Evans (1930) and Barnard (1930)

In Roesler s series the average age at death was 36 with an extreme of 75

When mitral stenosis is present the lesion is well tolerated and some cases attain a remarkable age Lutembacher s case had seven pregnancies and reached the age of 61 whilst Firket s case experienced eleven pregnancies and attained the age of 74 The average age at death in McGinn and White s series was 35 years

It may be remarked here that the longevity of the average case of the Lutembacher syndrome has led to a surgical approach in the treatment of mitral stenosis The subjects of the Lutembacher syndrome are not prone to the inconveniences and disasters of uncomplicated mitral stenosis They are exempt from acute pulmonary oedema and haemoptysis and die of right heart failure and not of pulmonary congestion It appears that a defect of the auricular septum affords protection from a mounting pressure in the left auricle It is therefore possible that some cases at least of mitral stenosis might achieve benefit from the creation of an auricular septal defect or its equivalent The effect of a septal defect can be secured with little difficulty by anastomosis between a left superior pulmonary vein and the azygos vein using a Blakemore tube This operation has already been successfully accomplished

ANEURYSM OF THE TRUNK AND THE MAIN BRANCHES OF THE PULMONARY ARTERY

Extreme dilatation of the pulmonary artery is not uncommon in a variety of conditions particularly when observed by the X ray and has frequently led to an erroneous interpretation of congenital heart disease Assmann (1928) state that one third to one-half of congenital cardiac cases have enlargement of the pulmonary artery and about

one fifth of these cases are of patent ductus arteriosus Perry (1931) found prominence of the pulmonary conus in 26 8 per cent of his cases of congenital heart disea e Lincoln and Spillman (1928) found similar prominence in 21 3 per cent of normal children In passing it may be mentioned that rheumatism of the pulmonary artery may cause marked prominence of the pulmonary conus as in the case of Schwartz and Schelling (1931) Roesler (1936) found about half the cases of pulmonary aneurysm to be associated with congenital heart or vessel lesions Boyd and McGavack (1939) in an analysis of 111 recorded cases of pulmonary aneurysm (examples of simple diffuse dilatation being excluded) found accompanying congenital lesions in 66 per cent and these lesions were considered to be important predisposing factors in the development of an aneurysm in 43 2 per cent In their analysis in discussing the role of other precipitating factors which might initiate the aneurysmal process they found active syphilis in 31 7 per cent arteriosclerosis in 23 per cent and other infections in 17 2 per cent The congenital lesions most often present are defects of the inter auricular septum patent ductus arteriosus and pulmonary stenosis A bicuspid pulmonary valve has been recorded as associated and it is interesting with its possible implication of unequal division of the truncus Sometimes an infective arteritis of the pulmonary artery may be an important factor Similar views are shared by Deterling and Clagett (1947) The anatomical features of the pulmonary artery that allow its dilatation are its relatively thin walls in comparison with other arteries of similar calibre its deficiency of muscular and elastic tissue in the medial coat especially along the relatively unsupported anterior and lateral walls of the trunk and its low tonus in common with all vessels in the pulmonary circuit These factors render its distension a comparatively easy matter in the presence of a shunt of the magnitude that occurs in a defect of the interauricular septum or in a patent ductus arteriosus The intervention of infection renders dilatation all the more likely

It is important but difficult to determine what is an aneurysm as distinct from mere dilatation of the pulmonary artery Any student of the literature soon becomes aware that many of the described aneurysms have begun as simple fusiform dilatation and then with the develop ment of arteriosclerosis have become aneurysms Boyd and McGavak would only include as aneurysms those cases in which there is post mortem evidence of more or less circumscribed dilatation of the pulmonary artery with some degeneration of the vessel wall This by definition almost excludes the possibility of accurate diagnosis during life in a great many cases Aneurysmal dilatation of the pulmonary artery a term quite commonly employed would continue to get over some of the difficulty

As regards the site of the aneurysm it may involve the trunk or one

or both branches of the pulmonary artery The branches alone may be involved to the exclusion of the main trunk

Cases associated with a pulmonary stenosis are those of Mantovani (1902) Arnheim (1905) and Botenga (1936) and in these the trunk alone was involved Arnheim s case exhibited valvular stenosis It is at present a matter of difficulty to adequately explain post stenotic dilatation Cases in which the ductus was patent and these appear to be the commonest include those of Terplan (1934) where there was coincident infection Joules (1934) where in addition to a patent ductus there was a defect both of the auricular and ventricular septum and the trunk and both branches were involved and Scott (1934) where there was a moderate coarctation Recent cases are those of Palmer and Kempf (1939) Johannsen and Connor (1943) and Hartwell and Tilden (1943) The auricular septal defect figures largely in the case of Menzini (1935) where the right branch was involved Involvement of the right branch is not uncommon and is readily recognized by its comma radiological picture The work of Bedford Papp and Parkinson (1941) has indicated that dilatation of the trunk and branches is so common and marked in the auricular septal defect as to raise the question as to what degree of enlargement may truly be classed as aneurysmal Wilkinson s (1940) case of massive enlargement of the pulmonary artery showed only a small interventricular septal defect In this case both trunk and branches were involved

Despite the frequency of the auricular septal defect and the patent ductus arteriosus in the female the distribution of cases between the sexes in congenital cases shows only a slight preponderance in the female It also appears that there are two main peaks of incidence the first from birth to the twenty ninth year representing those cases in which a congenital anomaly forms the basis of aneurysm formation and from the fortieth to fifty ninth year where such factors as syphilis and arteriosclerosis become operative

CLINICAL FEATURES Aneurysm of the trunk of the pulmonary artery either alone or combined gives rise to physical signs and symptoms whereas aneurysm of a branch tends to give rise to symptoms alone In addition there are the signs and symptoms of an accompanying congenital heart lesion Boyd and McGavack (1941) state that in aneurysm involving the trunk there is stasis in the lesser circulation characterized by dyspnoea cyanosis fullness in the chest and perhaps haemopytsis There may be prominence of the chest especially in the region of the second and third left costal cartilages Pulsation is often visible in the second left space and at the same site there may be a thrill and a loud superficial systolic murmur Hypertrophy and dilatation of the right side of the heart are present with corresponding changes in the electrocardiogram and a prominent pulmonary conus in the radiological picture A cough is common in

both aneurysm of the trunk or pulmonary branches and it has been designated as brassy by Foster (1920) It is due either to irritation of the recurrent nerve to bronchial irritation or to pulmonary thrombosis in which latter case it is accompanied by blood stained sputum Dyspnoea and cyanosis appear earliest in congenital cases Peripheral oedema is rare When failure does occur it is of the right sided type with a large and perhaps pulsating liver from tricuspid incompetence ascites and peripheral oedema

Some of the principal clinical features are illustrated by the cases recorded below In Sondergaard s (1936) case a male of 31 there was cyanosis and clubbing of sixteen years duration together with dyspnoea on exertion and occasional haemoptysis The heart was slightly enlarged and there was bulging of the preacordium A blowing systolic murmur was present The radiogram showed a small aorta with a greatly enlarged pulmonary artery the hilar arteries being greatly increased Autopsy showed a widely patent foramen ovale A case reported by Bedford (1928) a female aged 36 exhibited breathlessness on exertion and pain of anginal type The heart was enlarged and there was a thrill and pulsation of expansile type in the second and third spaces A systolic murmur was present over the pulmonary area and occasionally a diastolic murmur of pulmonary incompetence was heard She developed cyanosis and had occasional haemoptysis The symptoms and signs were attributed to a patent ductus arteriosus and infective arteritis of the pulmonary artery Moench s (1924) case a female aged 29 showed few symptoms except vertigo and breathlessness and died suddenly There was a large patent ductus arteriosus admitting the index finger a bicuspid pulmonary artery and a linear rupture of the greatly dilated pulmonary trunk Wilkinson s (1940) case a girl of 13 suffered with breathlessness She had a grossly enlarged heart and an enormous pulmonary artery Dullness extended up to the second left space in the midclavicular line There was a long systolic murmur in the second left space the pulmonary second sound was well heard and there was a diastolic murmur She died suddenly from rupture of the dilated pulmonary artery which showed a linear split about two inches in length one inch above the pulmonary valve The pulmonary valve was bicuspid and there was a small defect at the base of the interventricular septum Joules (1934) reporting a case of persistent ostium secundum in a female aged 37 states that physical signs are scanty until pulmonary incompetence develops Cyanosis is often late in appearance and the presence of a heart lesion may not be recognized until late in adult life Favorite s (1934) case of cor biatriatum triloculare exhibited a greatly dilated pulmonary artery which ruptured and caused death In Wahl and Gard s (1931) case a female of 19 an aneurysm of the right branch led to a diagnosis of mediastinal tumour At operation there was fatal incision of an aneurysm The foramen

ovale was patent and measured 22 mm in diameter Slight cyanosis and clubbing were present Henschen (1906) who analysed forty cases found the main symptoms to be dyspnoea some degree of cyanosis and occasional haemoptysis There may be substernal pain (cf aneurysm of the aorta) either over the site of the dilated pulmonary artery or provoked by exertion and of definite anginal type The physical si ns were generally bulging and pulsation in the second and third left intercostal spaces The heart was enlarged to left and right Dullness corresponding to the dilated vessel was present in the second and third left spaces The auscultatory signs were a systolic murmur often of a rather superficial character and occasionally a diastolic murmur was present A thrill was frequent in the pulmonary area The usual signs of an aneurysm tended to be absent although there may rarely be paralysis of the recurrent laryngeal nerve

RADIOLOGY The picture is essentially similar to that of the inter auricular septal defect There is a large actively pulsating pulmonary artery and increased depth and breadth of the hilar shadows especially on the right where the pulmonary branch is unobscured by the cardiac shadow There is in addition indentation of the oesophagus below the aortic impression in the right anterior oblique position Perhaps one of the best examples extant is that of Wilkinson (1940) The pulmonary origin of ah aneurysm has been demonstrated by angiocardiography by Robb and Steinberg (1940) and others

ELECTROCARDIOGRAM An aneurysm of the pulmonary artery like the similar condition of the aorta is *per se* unlikely to cause an alteration on the electrocardiogram unless there is associated pathology in the pulmonary valve conus or elsewhere Because the associated congenital abnormality is either a defect of the interauricular septum or a patent ductus arteriosus both of which allow a shunt from left to right and so over burdening the pulmonary circulation right axis deviation is usual if not inevitable

DIFFERENTIAL DIAGNOSIS This may be a matter of considerable difficulty in some cases An aortic aneurysm involving the descending aorta may usually be identified on radiological grounds and by the absence of enlargement of the pulmonary branches A patent ductus arteriosus usually presents the classical continuous murmur which is characteristic If however thrombosis occurs in the ductus it may be impossible to differentiate it from a pulmonary artery aneurysm Inter auricular septal defects are generally recognized by the absence of distinctive murmurs the association of mitral valve lesions and the absence of a thrill and chest deformity which are frequent in aneurysm Mediastinal tumours may be recognized by kymographic methods

COURSE AND PROGNOSIS Aneurysmal dilatation of the pulmonary artery may be compatible with life until the third decade when symptoms are apt to occur Boyd and McGavack gave the average age at

death as 32 years Death usually results either from rupture of the pulmonary artery intercurrent infection bacterial endocarditis or intercurrent disease Tuberculosis claims some victims

Congenital Dilatation of the Pulmonary Artery and its Branches

(Idiopathic Dilatation of the Pulmonary Artery)

As an isolated abnormality unaccompanied by any other congenital abnormality congenital dilatation of the pulmonary artery and its branches is a rare condition Considerable confusion exists in relation to this anomaly which by definition must exclude those cases where there is a patent ductus arteriosus interauricular septal defect or evidence of rheumatic or syphilitic disease of the heart blood vessels or lungs The difficulty arises because these latter conditions like isolated dilatation of the pulmonary artery may be acompanied by sclerotic changes in the pulmonary artery and by a relatively small or hypoplastic aorta

The case of Zuber (1904) from which originated the term congenital dilatation of the pulmonary artery is thus excluded because a patent ductus arteriosus was present Oppenheimer (1933) reported several cases with two post mortems both of which showed a dilated trunk and branches of the pulmonary artery together with hypertrophy of the right ventricle and a small aorta The most convincing evidence in favour of a congenital origin of the condition is the presence of a hypoplastic aorta although it is of course possible that cases without aortic hypoplasia may equally have a congenital origin Assmann (1929) was perhaps the first to suggest that unequal division of the truncus arteriosus was the primary event This view has been supported by the French cardiologists notably Laubry *et al* (1941) who describe a condition of grosse pulmonaire petite aorte although but one of their reported cases is wholly acceptable as being of the type under discussion Similarly one of the cases of Kourilsky *et al* (1941) might also be acceptable in this category By far the greater number of cases where there is dilatation of the pulmonary artery and a small aorta are associated with an interauricular septal defect and it seems that this unequal division of the truncus arteriosus may result not only in auricular septal defects but also in isolated dilatation of the pulmonary artery

There is also a group of cases reported as primary pulmonary arteriosclerosis defined by Brenner (1935) as being sclerosis of the pulmonary *artery with right sided hypertrophy unaccompanied by any other heart* or pulmonary disease East (1940) and De Navasquez Forbes and Holling (1940) also describe cases with dilated pulmonary arteries and right sided hypertrophy associated with pulmonary arteriosclerosis

and attributed to pulmonary hypertension of unknown origin There might be some reasonable criticism of this view There is experimental evidence that hypertension of the pulmonary artery may cause arterio sclerosis but there is no clear knowledge that pulmonary arterio sclerosis of itself can result in pulmonary hypertension In mitral stenosis for example notable arteriosclerosis may be present in the pulmonary trunk and branches without great dilatation of the arteries despite the presence of pulmonary hypertension suggesting that if neither arteriosclerosis or hypertension cause dilatation then there might well be some additional congenital structural factor to account for it From this one might well infer that a congenital unequal division of the truncus is a probable etiological factor

On the assumption of a congenital disproportion in size as between the aorta and pulmonary artery Gold (1946) advances the ingenious theory that stress in the dilated pulmonary artery is greater than in a vessel of normal calibre Thus when there is a column of fluid at constant pressure in a cylinder the stress at any point on the cylinder wall is directly proportional to the diameter of the cylinder at that point From this in a dilated vessel the stress is greater than it would be if the vessel were normal in diameter Accepting the hypothesis that arteriosclerosis is a degenerative lesion initiated by increased stress on the vessel wall then sclerotic changes would readily appear in the dilated vessel These changes appear in the absence of hypertension but extension to the medium and smaller branches of the pulmonary artery with thickening of their walls and narrowing of their lumen must ultimately lead to hypertension and with it right ventricular enlarge ment and finally failure

No mention has been made of Ayerza s disease an ill defined syndrome with cyanosis and pulmonary vascular sclerosis In the welter of cases reported as Ayerza s disease one finds pulmonary arterio sclerosis considered by some to be secondary to pulmonary or bronchial disease and by others to be due to syphilitic arteritis of the pulmonary vessels Undoubtedly the reported cases show a dilated pulmonary artery and right ventricular hypertrophy In a few cases mention is made of hypoplasia of the aorta It seems that there is grave doubts as to the specificity of the Ayerza syndrome and some of the reported cases may be true examples of congenital dilatation of the pulmonary artery

CLINICAL PICTURE There are no characteristic symptoms or physical signs to reveal the presence of this abnormality which is often first recognized on radiological examination Cyanosis is usually slight and may be noted in regard to the lips or finger tips Some breathlessness on exertion is common The heart is enlarged to the right A very few cases have presented a basal systolic murmur and thrill In most cases an accentuated pulmonary second sound has been noted Moderate

polycythaemia may occur The electrocardiogram shows a right axis The X ray picture confirms the hypertrophy of the right ventricle and shows a greatly enlarged pulmonary artery It can be said that the general picture is that of a subacute cor pulmonale in which the only constant discoverable abnormalities are dilatation of the pulmonary artery an accentuated pulmonary second sound and right axis deviation in the electrocardiogram Apart from the condition under discussion pulmonary arteritis and lymphangitic carcinomatosis of the lungs the latter secondary to a symptomless carcinoma of the stomach or breast should be considered in the differential diagnosis Cardiac catheterization may be necessary to make a diagnosis from an auricular septal defect

Course and Prognosis Apart from some dyspnoea on exertion the condition may remain benign and latent until the enlarged right ventricle begins to fail Failure once established does not respond well to treatment Death may occur at any age but usually in the second to fourth decade

Isolated Dilatation of a Branch of the Pulmonary Artery

Soulie Bourrain and Joly (1943) have drawn attention to dilatation of a branch of the pulmonary artery most often the left The aorta is normal in size and cases therefore do not correctly fall into the group of unequal division of the truncus arteriosus Soulié *et al* present six cases five involving the left branch One case only had slight cyanosis and clubbing A systolic murmur was audible in the second left space and also in the left interscapular region Some degree of right axis was present in the electrocardiogram The X ray picture showed enlargement of the left pulmonary branch and no enlargement of the pulmonary trunk Unfortunately there are no post mortem examinations In other reported cases the right and left branches have been dilated with the trunk of the artery apparently of normal size These cases may or may not be accompanied by right axis in the electrocardiogram but the published X ray pictures show enlargement of the heart An example is the case of Lenegre Roudinesco and Marquis (1943) As a result of this work a personal search for these cases revealed several examples but no post mortem material is forthcoming It may be remarked that in the isolated patent ductus arteriosus the left pulmonary artery may be larger than the right due to the direction of the stream of blood from the ductus largely into the left pulmonary artery

CHAPTER XI

MALADIE DE ROGER
ISOLATED DEFECTS OF THE VENTRICULAR SEPTUM
CONGENITAL HEART BLOCK

Roger (1879) was the first to describe a congenital defect of the heart in which symptoms were absent although *definite constant physical signs were present* Dupre (1891) verified a case at autopsy and named the syndrome the maladie de Roger

It is possibly the commonest of all congenital abnormalities but the condition is so benign and symptomless that many cases escape observation or are designated by some other diagnosis Perry (1931) reviewing the congenital heart cases of a school cardiac clinic found that 35 per cent were referable to this category Muir and Brown (1934) analysing similar material found that 37 per cent of cases presented the signs of an isolated interventricular septal defect

Defects of the ventricular septum are generally situated at the base of the septum but may rarely be present at other sites Entire absence of the ventricular septum cor biatriatum triloculare is considered *elsewhere (Chapter XV) Rokitansky (1875) divided the septum into* two parts an anterior and a posterior with the membranous septum as the boundary between them The anterior part of the anterior septum is derived from the bulbus cordis The rest of the septum is evolved from the septum inferius and the fused cushions of the atrial canal *Defects of the anterior* (bulbar) septum are rare The common position of the isolated septal defect is just anterior to the membranous septum beneath the aortic cusps Defects may be found in other regions of the ventricular septum as for example in the case of Weiss (1927) where a large interventricular communication existed near the apex

The interpretation of an isolated defect as a persistent interventricular foramen is questionable Fraser (1917) has shown that the interventricular foramen becomes the connecting link between the aorta and the outflow part of the left ventricle The relation of defects in this region to the evolution of the bulbus cordis is evident from the frequency with which a *septal defect is associated with lesions of the* pulmonary tract Laubry and Pezzi (1923) have pleaded for caution in the diagnosis of an isolated defect in view of *this association* Keith (1909) and Abbott (1927) consider that isolated defects of the ventricular *septum* anterior to the membranous septum are due to a primary arrest of growth of unknown origin It may be very difficult to separate

some cases from the Eisenmenger complex with slight dextroposition of the aorta a minimal form of transposition of the vessels

ANATOMY In appearance the defect is variable in size admitting only a probe in many cases but occasionally large enough to admit a little finger (fig 53) Circular or oval in shape the margin of the small defect is generally surrounded with fibrous tissue affording a marked contrast to the fleshy margin of the larger defect in graver anomalies such as the tetralogy of Fallot *Defects in the common situation* that is in the posterior part of the anterior septum usually open into the right ventricle beneath the septal cusp of the tricuspid valve or occasionally into the right auricle above the line of attachment of the same cusp Defects in the anterior part of the anterior septum open into the infundibulum of the right ventricle and are usually associated with anomalies of the pulmonary tract and dextroposition of the aorta (see Tetralogy of Fallot)

The morphological implications of ventricular septal defects are of interest Bulbar septal defects correspond to the anterior communication between the dorsal and ventral ventricles of the turtle s heart *Defects anterior to the membranous septum are the homologue of the* foramen between the aortic ventricles of the turtle (Abbott and Shanly 1922) Defects of the lower or muscular part of the septum are rare and correspond to the communication between the dorsal and ventral ventricles found normally in the heart of the python (Weiss 1927 Mason and Hunter 1937)

A defect of the interventricular septum may be an acquired condition It may be the result of perforation of a gumma of the septum or be the sequel of an infarct of the septum subsequent to coronary disease

The presence of a shunt from left to right through the defect is *shown by the occurrence of an area of fibrosis on the wall of the right* ventricle opposite the defect Similarly in the event of infective endocarditis involving the orifice of the defect vegetations may be found on the opposite wall of the ventricle or on the tricuspid leaflet

There is some evidence that an interventricular septal defect large and important in infancy may with growth of the heart become relatively small and unimportant in comparison with the size of the heart It may ultimately become negligible and fail to give rise to physical signs It is also possible as Parkes Weber (1918) suggests that the fibrous tissue surrounding the defect may contract and obliterate the orifice Cases have been recorded with disappearance of both thrill and murmur by Parkes Weber (1918) French (1918) *Stamm (1918)* Still (1924) Muir and Brown (1937) and Perry (1937) Unfortunately there is no post mortem evidence to corroborate these observations Fibrosis in the region of the defect may involve an aortic cusp and lead to its deformity and consequent aortic incompetence (Brandeburg

1934) (fig 54) In the unique case of Allen (1941) the right aortic cusp herniated through the defect and adhered to the anterior pulmonary cusp

CLINICAL PICTURE There are no symptoms that can be attributed to this abnormality Its characteristic feature is an absence of symptoms with marked physical signs The description of Roger (1879) may well be quoted In his conclusions he finds that

There is a developmental defect of the heart from which cyanosis

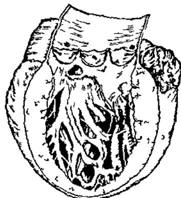


FIG 53

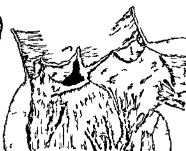


FIG 54

FIG 53 The maladie de Roger in a child who died suddenly
FIG 54 Maladie de Roger with involvement of an aortic cusp in fibrous tissue originating from the defect and causing aortic incompetence

does not result despite a communication between the two ventricular cavities It consists of an opening in the interventricular septum It is revealed only on auscultation by a physical sign with a very definite character this is a long loud murmur It begins in systole and is prolonged to such an extent as to entirely cover the natural tic tac of the heart sounds It has its maximum intensity not at the apex but over the upper third of the praecordial region It is chiefly medial in position like the septum itself and from this central position it diminishes in intensity uniformly as one moves the stethoscope over the chest It coincides with no other sign of heart disease except a harsh thrill which accompanies it This murmur is the pathognomonic sign of an interventricular septal defect

Little may be added to this original description of Roger which is all the more remarkable because his observations were unconfirmed by post mortem evidence until eleven years later The mesocardial murmur occupying the whole of systole and occasionally prolonged into diastole is of maximum intensity in the third and fourth left spaces close to the

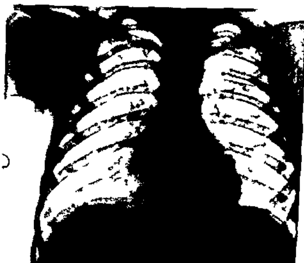
sternum It may frequently be heard in the interscapular region In about two thirds of the cases a thrill corresponds in site to the murmur Some cases have been described as permanently cyanotic but these are not cases of the true maladie de Roger where cyanosis is only a transient phenomenon and never established Cyanosis when it occurs is of the type of cyanose tardive and is due to a reversal of flow through the defect under conditions of stress such as pulmonary infection or as a terminal event It would demand a defect of sufficient size to allow an important shunt Experiments with a spirometer have failed to induce cyanosis There is no shortness of breath on exertion Any symptoms that are present are generally traceable to the strong functional element that pervades these subjects The very striking nature of the physical signs may give rise to misgivings on the part of the physician and inadequate appreciation of the lesion present may result in the imposition of much unnecessary restriction of activity A small defect may give rise to more marked signs than a large one because conditions are favourable for the production of a murmur in the small defect A large defect may give rise to neither bruit nor thrill

Radiological Picture A globular heart silhouette has been frequently described as characteristic of the condition This picture is rare and generally there is no alteration in the shape or size of the heart shadow There may be some slight enlargement of the heart as a whole with a tendency towards the assumption of a globular form in cases where there is a large septal defect The pulmonary artery may be slightly dilated imparting a straight left border to the heart (fig 55) Considerable enlargement of the pulmonary artery and its branches has occasionally been described but this is exceptional and may be ascribed to structural changes in the pulmonary artery or to an associated patent ductus arteriosus

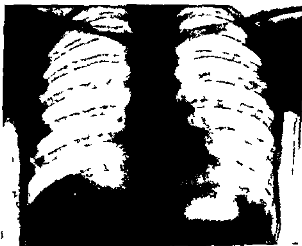
It is of course possible that a large shunt of blood from left to right through the defect may ultimately result in enlargement of the right ventricle and pulmonary artery The occasional occurrence of arteriosclerosis in the pulmonary artery may be evidence of this increased strain upon the pulmonary circulation Angiocardiography is of little use to demonstrate an isolated septal defect

Electrocardiogram The electrocardiogram is generally physiological There may be changes in the axis deviation especially to the left (fig 56) A right axis occurred in a case of the author in which the defect was at the apex of the septum Conduction defects may be present with widening of the QRS or a prolonged PR interval These are uncommon Complete heart block is discussed below

Course and Prognosis The condition may be entirely latent and only discovered at school medical or life insurance examination The subjects are exposed to the risks of an infective endocarditis and in Abbott s (1931) series of fifty cases thirteen died of bacterial



Skiagram of typical case showing a globular contour



Typical case with a straight left border

Fig 55

endocarditis In Gelfman and Levine s (1942) series of autopsied cases the incidence of bacterial endocarditis was 23 per cent in patients of all ages and 57 per cent in patients over two years of age The infective process may but does not always involve the margins of the defect Mural thrombi may be present on the wall of the right ventricle where the abnormal shunt impinges opposite to the defect The aortic and tricuspid valves may be involved by direct spread Excellent examples are those of Perry (1936) Falconer (1937) Eigen and Abel (1941) and

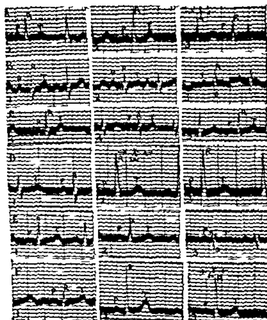


Fig 56 The electrocardiogram in six cases of the maladie de Roger

Zeman (1945) the latter in a case aged 61 The case of Allen (1941) is interesting because the right aortic cusp herniated through the defect and fused with the anterior pulmonary cusp giving a picture of aortic incompetence and pulmonary stenosis Audibert (1933) thought that the isolated interventricular septal defect was rarely involved in bacterial endocarditis Post mortem statistics have their fallacies particularly in a latent abnormality such as the maladie de Roger but unquestionably bacterial endocarditis is a serious risk Gelfman and Levine (1942) found the highest incid nce of bacterial endocarditis with this defect The presence of congenital heart block does not necessarily seriously influence the prognosis in these cases

Normally the maladie de Roger is quite compatible with a vigorous and active life and should be no handicap in most employments No treatment is required and no restrictions should be imposed The average age at death in Abbott s series was 14 years with an extreme of 49 Weiss s case lived to the age of 79 Personal cases have been under observation for eighteen years and show no deterioration

DIAGNOSIS Diagnosis depends upon the presence of the typical physical signs of mesocardial systolic murmur and often a thrill in a patient with no symptoms Great difficulties may arise in early child hood where it may be impossible to differentiate a patent ductus arteriosus or even an auricular septal defect until the age of five or six when more characteristic physical signs may be evident A ductus has the typical Gibson murmur by this age and the auricular septal defect a right axis in the electrocardiogram with a prominent conus and pulmonary branches A functional murmur may be very loud but is so rarely heard in the back that audibility in the back is good evidence of its organicity Phonocardiographic methods may well separate the organic from the functional bruit Some help may be gained from X ray examination Angiocardiography is undoubtedly useful but not without its risks Lastly intracardiac catheterization will demonstrate the presence of a left to right shunt It seems doubtful if these two latter investigations have any serious place in the diagnosis other than academic interest in a lesion that is so generally benign and well tolerated

VENTRICULAR SEPTAL DEFECT ASSOCIATED WITH PULMONARY HYPER TENSION The only reported case is that of Burchell *et al* (1948) The physical signs were typical of the conventional ventricular septal defect The electrocardiogram showed a right axis and the radiological examination a slight enlargement of the heart with prominent pul monary conus and large hilar shadows In effect the total picture was indistinguishable from the Eisenmenger complex Identification of this particular case was established by catheter technique There was evi dence of a large left to right shunt pulmonary hypertension and a normal oxygen saturation remaining normal with exercise

ANEURYSM OF THE VENTRICULAR SEPTUM Aneurysm of the ventricular septum involves the membranous septum The direction of the aneurys mal pouch varies and consequently when viewed from the right ventricle the sac may appear beneath the septal cusp of the tricuspid valve if the direction is downwards or on the auricular margin of the same cusp if directed upwards The aneurysm is a small sac shaped structure with a circular mouth of about 1 cm diameter It does not however always conform to this type for the lesion described by Guccione (1926) was funnel shaped The depth varies and is usually from 1 to 2 cm Generally there are other congenital anomalies present including displacement of the coronary arteries indicative of slight

transposition Subaortic stenosis was present in the case of Rae (1936) In the two hearts of Lev and Saphir (1936) both derived from mongols there was a large patent foramen ovale and fenestrations of the septum primum These same authors state that since the first case was described by Laennec in 1826 about seventy cases have been reported A patent foramen ovale appears to be frequent and in Merkel s (1869) case there was a defect of the ventricular septum The auricular septum was absent in the case of Zadoc Kahn and Cousin (1925) Calcification may occur in the aneurysmal sac

PATHOGENESIS A number of theories have been advanced in explanation of the formation of such an aneurysm Endocarditis was offered as an explanation by Rokitansky (1875) and by Merkel It appears most likely that the explanation is purely embryological and is the result of malformation of the membranous septum owing to a small degree of transposition of the aorta and failure of the inter ventricular septum to move to the right Anatomical support to such a hypothesis has been adduced by Mall (1912) Eakin and Abbott (1933) and Bishop and Trubek (1936) and the displacement of the coronary orifices is confirmatory It is thus a detorsion defect Pressure in the left ventricle acting upon the malformation of the membranous septum results in aneurysm formation

There are no symptoms and the condition has never been recognized in life

CONGENITAL HEART BLOCK This interesting abnormality is frequently described as being associated with defects of the interventricular septum Scattered through the literature are a number of cases where heart block in young children has been accompanied by the signs of this defect The proximity of the bundle of His to the membranous septum and the supposition that defects of the interventricular septum involve the membranous septum has led to an easy and attractive hypothesis that the cause of congenital heart block is due to a defect of the interventricular septum If such a concept were true then congenital heart block would be a common clinical finding for the interventricular septal defect either alone or combined with other lesions is one of the most frequent of cardiac anomalies A review of forty four accepted cases of congenital heart block by Yater Lyon and McNabb (1933) showed that signs of a septal defect were present in twenty seven and these were confirmed by autopsy in five As will be shown later the site of the septal defect has a very important influence in relation to heart block It is an interesting observation that in the cyanotic case where the defect of the septum is of considerable magni tud heart block is very rare

Cases of congenital heart block should perhaps be divided into two categories Firstly those cases where the condition has existed since birth and due solely to the direct effects of the anatomical malforma

tion Secondly those cases of congenital heart disease which develop block subsequent to birth from strain or other pathological cause

Congenital complete block has been described in a variety of other abnormalities In Hoekenga s (1945) case there was a cor biatriatum triloculare with tricuspid and pulmonary atresia The ductus and foramen ovale were patent Atrial and ventricular septal defects were present in the case of Peel (1943) Dextrocardia accompanied the case of Leys (1943) The case of Jaleski and Morrison (1943) had two pregnancies

The rudiments of the conduction system appear at about the fifth week and at this time the auriculoventricular node may be identified in the wall of the lower posterior part of the common auricle Co incidently division of the atrial canal by fusion of the anterior and posterior endocardial cushions takes place so that the auriculo-ventricular orifices are formed Up to this time there has been con tinuity of the muscle fibres of the auricle and ventricle but this con tinuity is practically destroyed by ingrowth of the connective tissue of the atrial ring (annulus fibrosus) A few fibres situated posteriorly between the posterior endocardial cushions and the annulus fibrosus escape and these fibres after further differentiation become the bundle of His Monckeberg (1924) showed that in nearly all cases of septal defect from the smallest down to complete absence of the ventricular septum the bundle was conserved although its position might be abnormal Thus in a complete absence of the ventricular septum the bundle may be found running along the tag of the rudimentary septum and even in the cor biloculare it exists on the posterior wall of the common ventricle and may be observed to divide into right and left branches The work of Flack and Mall (1911) indicates that the inter ventricular septum is not formed by the growing up of the septum but by hollowing out of the embryonic muscle to form the ventricles In their opinion the tip of the septum inferius represents the inner wall of the primitive cardiac tube and thus the bundle persists at this point This accords with the observed facts because in septal defects the bundle is found streaming over the posterior part of the defect to reach each ventricle

Abbott (1927) states that defects are most often found at the base of the septum anterior to the membranous septum The normal position of the bundle is in the posterior part of the membranous septum which it traverses to become the left branch If an isolated septal defect is accepted as one of the most frequent of congenital cardiac abnormalities it will readily be appreciated that as the defect is anterior to the membranous septum involvement of the bundle is most unlikely

Consideration of the above data suggests that the most vulnerable portion of the developing bundle is that part of it which lies in the posterior part of the endocardial canal between the endocardial

cushions This region of the atrial canal is a key point in the developing heart for from the endocardial cushions the auriculoventricular orifices and cusps are derived and further the endocardial cushions play an important part in the formation of the auricular ventricular and membranous septa Thus such lesions as a persistent ostium primum an absent auricular septum a common auriculoventricular orifice or a biloculate heart might reasonably be expected to be accompanied by disturbances of conduction if not actual block Likewise the bundle might be interrupted at this point by abnormal ingrowth of the fibrous tissue of the annulus fibrosus in the absence of any gross septal or other abnormality Conceivably the bundle might be involved in an infective process Lastly it should not be overlooked that tumours especially lymphangiomata have been described as causes of block

It is natural to inquire how far these ideas are supported by the observed facts It had been held by some with considerable justification that as the bundle is developed in advance of the septa defects of the septa *per se* are unlikely to be the cause of heart block Monckeberg (1924) states that even in the most complex and severe malformations it is unusual for the conduction system to be involved The bundle can be identified in these cases although its site may be abnormal He also remarks that where a persistent foramen primum is present the auriculoventricular node is displaced from its usual position but he never observed a case in which block was present Mahaim (1931) considers that the persistent ostium primum places the bundle in an abnormal posterior position and thereby increases its vulnerability owing to the anomalous changes taking place in the endocardial cushions Clinical substantiation comes from the cases of Yater Leaman and Cornell (1934) and Wallgren and Winblad (1937) where a persistent ostium primum was associated with complete block In both cases the actual cause of block was interruption of the bundle by ingrowth of fibrous tissue from the atrial ring

If the membranous septum is absent some disturbance of its near relation the bundle might be expected Heart block was found in absence of the membranous septum by Perotti (1928) Wilson and Grant (1926) and Yater Lyon and McNabb (1933) Cases in which the septa have been intact are those of Yater (1929) McLellan (1935) and Wallgren and Winblad (1937) In all of these cases it is presumed that the bundle was interrupted by fibrous tissue ingrowth and microscopical evidence confirms this in Yater s and Wallgren and Winblad s cases

Signs of ventricular septal defect have been present in fifty one of the seventy seven reported cases and in five of the twelve cases that have been examined at post mortem The defect has been large or complete in the cases of Moxon (1879) Wilson and Grant (1926) Abbott and Moffatt (1930) Yater Lyon and McNabb (1933)

Heart block can occur in other forms of congenital heart disease where the septum is *intact* Thus *in the case of Wadman (1945)* there was a patent ductus arteriosus and no evidence of other lesion Syncopal attacks occurred after exertion and an electrocardiogram showed complete block with a rate of 39 In the intervals there was a normal rhythm Leech (1930) also reported a case of ductus with complete block but the possibility of an interventricular septal defect was not wholly excluded It seems possible that variations in pressure on the *bundle or of tension in the bundle* may produce block Such changes in pressure on the bundle were thought by Nissé (1928) to be the cause

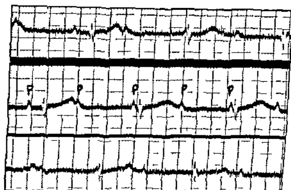
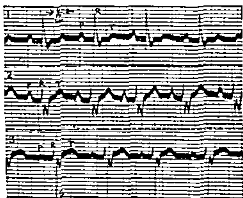


FIG 57 Congenital heart block Female aged 4 Physical signs of a ventricular septal defect

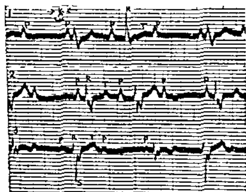
of alternating 2 1 and complete block in a congenital heart case with an interventricular septal defect Wilson and Grant (1926) showed the bundle to be divided into fine strands by the penetration of fibrous tissue in a case of 2 1 block and thought that these fine strands might easily be subjected to unusual pressure during failure Other cases such as those of Smith (1921) where block was absent on deep expiration suggests that the heart being then more horizontal tension on the bundle was decreased allowing the more ready passage of impulses In all probability as Wadman suggests the occurrence of convulsive *seizures or epileptiform* attacks may possibly in some cases be due to transient episodes of block Sudden strains or alterations in pressure relations on the bundle may cause block and if these strains are permanent the congenital heart case may actually acquire block In the case of auricular septal defect observed over a period of years a gradual lengthening of the PR interval and a prolongation of the QRS is sometimes observed

CLINICAL PICTURE Before a case is recognized as one of congenital heart block certain criteria enunciated by Yater should be satisfied

These are a slow pulse at an early age signs of congenital heart disease an absence of any history of infection especially diphtheria rheumatism or syphilis proof by graphic methods The occurrence of Stokes Adams attacks at an early age is good evidence These are however rare A



A



B

FIGS 58 A and B Electrocardiograms of a case aged 12 with signs of a septal defect and mental deficiency who developed heart block intermittently during observation

slow pulse may be present but heart block in childhood may be peculiar in that complete dissociation may exist with a comparatively rapid rate The earliest diagnosis of congenital heart block was made two weeks before birth in the case of Yater (1929) and Geiger and Hines (1940) give a table of the reported cases of pre natal diagnosis

There may be no symptoms directly attributable to the heart and occurrence of cyanosis is rare The heart may not be enlarged The physical signs are those of the maladie de Roger a mesocardial systolic murmur and perhaps accompanying thrill Even if no murmur is present there may yet be a large septal defect The blood pressure is often raised the average systolic level being 143 with an extreme of 190 in the series of Campbell and Suzman (1934)

Electrocardiogram All degrees of auriculoventricular dissociation may be present either complete or partial (fig 57) The QRS is not widened Alternation between partial and complete block occurred in a personal case (fig 58) The rate may vary from 36 to 92 the latter rate recorded in an infant by Nicholson Shilman and Green (1929) In Campbell and Suzman s (1934) series the average rate of eight cases was 50 and the average age of electrocardiographic examination was 12 although a slow pulse was observed on the average at $3\frac{3}{4}$ years Diphtheria as is well known is the commonest cause of complete heart block in young children It does not often however persist Complete heart block is uncommon in rheumatism although partial block is frequent and may last a considerable time Every effort should thus be made to exclude these diseases The routine electrocardiogram of 200 cases of interventricular septal defect has not shown an example of complete or partial heart block (unpublished personal series)

Course and Prognosis The prognosis appears to be that of the congenital heart abnormality present rather than that of the actual block Death may occur suddenly in a syncopal attack The average age at death was 7 years with a maximum age of 29 (Abbott)

CHAPTER XII

CYANOSIS

The earliest mention of cyanosis is by Paracelsus who referred to the condition as icteritia celestina seu cyanea Little is then heard of cyanosis until the eighteenth century when cyanosis began to be explained on the basis of the anatomical changes found in abnormal hearts Senac (1749) makes no mention whatsoever of cyanosis or shunts although he was credited with these observations by Henri Gintrac (1872) Morgagni (1761) reporting the case of a girl of 14 with a congenital pulmonary stenosis suggested that venous stasis might be the cause of cyanosis the venous stasis being the result of obstruction in the pulmonary artery Sandifort (1777) states clearly in his description of a case now recognized as the tetralogy of Fallot the arterial aorta was springing from both ventricles and had to receive all the blood from both They who recall to their minds the natural construction and work of the heart who compare the same with the degeneration just described understand well what a change took place in the circulation of the blood what a change in the function of the lungs Sandifort was thus very near the fundamental facts of cyanosis Corvisart (1813) was firmly of the opinion that cyanosis in many cases arose from unnatural communications between the cavities of the heart which allowed admixture of arterial and venous bloods This however was not always so for in some cases communications existed without cyanosis Louis (1826) admitted the possibility of mix ture but thought the principal factors involved were obstructive lesions in the pulmonary tract It is understandable that controversy might exist Gintrac (1824) and Bouillaud (1831) ranged themselves on the side of admixture Stillé (1844) and Grisolle (1869) favoured venous stasis and obstructive pulmonary tract lesions The discovery of a difference in the blood gases as between arterial and venous blood by Claude Bernard (1859) ushered in a period when cyanosis was thought to be due to alterations in the gases and increased carbon dioxide content of the blood Polycythaemia in congenital heart disease was first noted by Krehl (1888) The classic work of Lundsgaard and van Slyke (1923) clarified all these issues and emphasized the comparative value of shunt and venous stasis as factors in cyanosis Taussig (1939) recognized the essential fact that in those cases with a single ventricle and diminutive outlet chamber if the pulmonary artery arises from the main chamber it is large and cyanosis is minimal if on the other

hand it arises from the diminutive chamber it is small and cyanosis is intense The factor of the volume of blood reaching the lungs for oxygenation is thus of paramount importance and utilization of this principle is the basis of the operation now performed on the cyanotic case (Blalock and Taussig 1945)

Cyanosis is a blue colour of the skin and mucous membranes due to the presence of reduced haemoglobin in the circulating blood It may be the obvious presenting symptom of congenital heart disease and its presence and degree of intensity to a large extent reflect the severity of the underlying cardiac malformation Cyanosis is most easily observed in those areas where the capillaries are most numerous and is thus first detectable in the mucous membranes of the mouth the lips ears and finger tips

Normally in health the blood has an oxygen capacity of about twenty volumes per cent that is to say the haemoglobin of the blood can combine with 20 cc of oxygen It is usual to find that the arterial blood is only 95 per cent saturated equivalent to an actual oxygen content of 19 volumes per cent An average reduction of haemoglobin of about 5 volumes per cent leads to the venous blood containing about 14 volumes per cent oxygen These findings may be expressed in terms of oxygen unsaturation and accordingly the unsaturation of arterial blood is 1 volume per cent and that of venous blood 6 volumes per cent The mean capillary unsaturation is normally about 3 5 volumes per cent and being calculated from the arithmetical mean of the arterial and venous unsaturation does not correspond exactly to the average unsaturation of the blood in its transit through the capillaries Lundsgaard and van Slyke (1923) found that cyanosis appears when the capillary oxygen unsaturation is between 6 and 7 volumes per cent As 1 cc oxygen combines with 0 75 gramme haemoglobin it would appear that the threshold of cyanosis corresponds to about 5 grams of reduced haemoglobin per 100 cc blood As normal blood contains about 15 grams haemoglobin per 100 cc it means that at least one third of the haemoglobin must be in a reduced state The occurrence of cyanosis is thus dependent upon the absolute amount of reduced haemoglobin in the blood the amount of oxygenated haemoglobin present being of little importance If the haemoglobin content of the blood is increased as in polycythaemia cyanosis occurs more readily for in these circumstances an identical percentage of unsaturation corresponds to a greater amount of reduced haemoglobin Similarly where there is less than 5 grams of haemoglobin per 100 cc blood as in severe anaemias it is practically impossible for cyanosis to occur

In congenital heart disease apart from the chemical changes discussed above there are certain important factors determined by the anomaly present that result in the development of cyanosis In its most

severe form transposition of the great vessels without an inter ventricular septal defect cyanosis is not due to an insufficiency of blood reaching the lungs for oxygenation but depends upon the amount of oxygenated blood reaching the systemic circulation Only a patent ductus arteriosus (in which case the direction of flow is reversed) or a ventricular septal defect will allow oxygenated blood to reach the systemic circulation hence the larger the shunt through these channels the less the cyanosis The greater number of cases of cyanosis

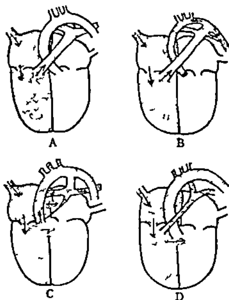


FIG 59 Origin of cyanosis in congenital heart disease
A normal B aortic atresia C pulmonary atresia
D Fallot

are due to an anatomical disposition or change in the vessels so that an inadequate volume of blood reaches the lungs for aeration or pul monary changes may be present which inhibit effective gaseous exchange in the alveoli To this may be added the effect of a venous arterial shunt where the aorta lies astride a high septal defect or arises wholly from the right ventricle or a common ventricle (fig 59)

In the patent ductus arteriosus or ventricular septal defect cyanosis may occur during severe pulmonary infection in failure or as a terminal event A reversal of the arteriovenous shunt occurs so that it becomes venous arterial Some have questioned the importance of a reversed shunt because dilatation of the skin capillaries and slowing of

the peripheral circulation have been demonstrated in these circumstances While the shunt may be adequate to produce cyanosis the local factors at the periphery may be *important in enhancing it*

Factors Productive of Cyanosis in Congenital Heart Disease

The all *important factor in the more severe types of congenital heart* disease surviving infancy is the volume of blood reaching the lungs for oxygenization This factor is obviously of great importance in such *conditions* as pulmonary stenosis or atresia with a high septal defect (tetralogy of Fallot) in cases where the pulmonary artery arises from a diminutive outlet chamber or in tricuspid atresia where there is often pulmonary artery hypoplasia or atresia In some cases the amount of blood reaching the lungs is adequate in the vegetative state of infancy but as the child develops and becomes active cyanosis appears and becomes established In most cases cyanosis is immediately apparent and may be extreme at an early age The significance of this factor has been stressed by the results of the Blalock Taussig operation the basis of which is to create an artificial ductus arteriosus and so increase the pulmonary circulation Even when this is successfully accomplished there must always be the factor of a venous arterial shunt directed through a ventricular septal defect with an overriding aorta which can never allow a normal oxygen saturation of the blood

In transposition of the vessels as mentioned above the lungs receive a full supply of blood but there is difficulty in returning the oxygenated blood to the systemic circulation In those cases where there is no patent ductus arteriosus or septal defect the blood simply recirculates to the lungs and the duration of life is brief cyanosis rapidly becoming extreme The presence of a patent ductus in which case flow in the *ductus is reversed from pulmonary artery to aorta* or of a ventricular septal defect allows oxygenated blood to reach the arterial circuit A large interventricular septal defect is therefore a favourable circumstance *and life has been prolonged thereby to a considerable age* A similar situation prevails in those cases where the pulmonary veins enter the right auricle or vena cava instead of the left auricle with the result that the oxygenated *blood recirculates in the pulmonary circuit*

In some cases of the Eisenmenger complex there appears to be difficulty in securing an adequate oxygenation of the blood despite an ample or even increased blood supply *It is probable that an increased* pulmonary pressure in evidence of which there is a hilar dance may cause changes in the alveoli sufficient to prevent adequate contact of the blood with oxygen Mention must be made of certain *contributory* factors which operate in the lungs Pulmonary disease whether infective or caused by pulmonary hypertension may lead to cyanosis A reduced vital capacity lowers gaseous exchange *Exercise by increasing the out* put of the heart may result in incomplete aeration of the blood and so

cause cyanosis or increase the cyanosis already present Polycythaemia by packing the alveolar capillaries may prevent adequate contact of all of the blood with the alveolar membrane

A venous arterial shunt is of importance and is nearly always present in the cyanotic case It occurs in those cases where there is a large septal defect with overriding aorta or where there is an absence of the inter ventricular septum In effect a variable volume of blood depending upon the position of the aorta the nature of the defect and the cardiac output is diverted into the systemic circulation instead of into the pulmonary circulation This will cause cyanosis not only by mixture of venous and arterial blood but also by leading to a less volume of blood reaching the lungs for oxygenation through a narrowed pulmonary tract The situation is well exemplified in the case of the single ventricle with the diminutive outlet chamber A small pulmonary artery rising from the diminutive chamber must necessarily result in cyanosis because of an inadequate volume of blood reaching the lungs and such blood is mixed blood because both auriculoventricular orifices enter the large single ventricle Conversely a pulmonary artery which is always large when it arises from the single ventricle and an aorta arising from the diminutive chamber does not lead to cyanosis until such time as other factors as an increased pulmonary pressure have produced their effects It has been calculated that 25 per cent or more of the circulating blood must be shunted to cause recognizable cyanosis

Mention may be made of those factors which further contribute to the production of cyanosis Peripheral stasis in the capillaries allows an increased utilization of oxygen and so causes an increased amount of reduced haemoglobin Further stasis in the capillaries implies dilatation of the capillaries which tends to make the distended capillaries more visible

Cyanosis in congenital heart disease is always manifest before clubbing of the fingers Clubbing is very rare in the first two years of life and is never present at birth It is proportional to the degree of cyanosis present Its earliest sign is thickening of the nail beds and gradually the nails and finger tips come to take on their characteristic drumstick appearance It is a curious observation that clubbing first appears in the index finger and thumb as also the changes in the un related condition of koilonychia (Lovibond 1938) Clubbing becomes extreme in long standing cases of cyanosis and may occasionally be associated with the changes in bone characteristic of the hypertrophic pulmonary osteoarthropathy of Pierre Marie (1890) Curiously enough secondary hypertrophic osteoarthropathy is apparently rare in con genital heart disease Locke (1915) in his review of 144 cases of hyper trophic osteoarthropathy ascribed only two to congenital heart disease In addition there are the cases of Shaw and Cooper (1907) and Means and Brown (1947) In this latter case a man of 26 blue from birth

there was regression of the signs of bone disease after an anastomotic operation

Lovibond states that the earliest change in clubbing is the develop ment of a firm transverse ridge distal to the last interphalangeal joint the skin being smooth and shiny and of a lilac hue The base of the nail gradually becomes raised and with it there is obliteration of the basal angle of the nail and finger At the same time increased curvature of the nail in both directions gradually develops Finally the tips of the fingers

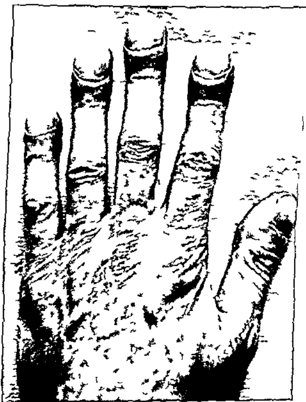


FIG 60 Clubbing of the fingers in congenital heart disease

become broadened and resemble a drumstick in appearance (fig 60) The causes of clubbing are related to a state of chronic passive conges tion of the capillaries of the nail bed a site where capillaries are par ticularly abundant and readily visible with the capillary microscope Oedema of the tissues between the nail bed and the bone then develops The accompanying anoxaemia and capillary dilatation together with

the slow circulation increased local deoxygenation and accum of metabolites all tend to produce clubbing

The eye changes are important There may be slight exophth The conjunctivae are suffused and congested and there may be haemorrhages in a few cases The retina has a darker shade than and the vessels are dilated and increased in number In sever the appearance of the vessels at their emergence from the opt is so characteristic that one might speak of retinal clubbing

Pathological effects of Cyanosis

POLYCYTHAEMIA The body gradually develops a remarkabl ance to oxygen unsaturation This is achieved by a compe polycythaemia which raises the oxygen carrying capacity of the and enables an adequate circulation within limits despite anal handicaps The stimulus to erythropoiesis is anoxaemia T cell count in the normal person is stated by Wintrobe and (1929) to be 5 800 000 In cyanotic congenital heart disease counts of seven and eight million are usual and the count ma as high as twelve million A high and rising count is of seriou nostic significance Coincidently with the increased number cells the haemoglobin is increased and the haematocrit level The polycythaemia of congenital heart disease is distinguishe that of Osler s disease by absence of enlargement of the spleen

Anoxaemia is not necessarily proportional to the severity of cy and can be very severe in infants before the development of any cyanosis or polycythaemia Anoxaemia is always accompar dyspnoea Dyspnoea with aggravation of cyanosis may occur v slightest exertion or it may occur in paroxysmal attacks prov crying feeding or even spontaneously Such attacks are more c in infants than in older children The attacks are spectacular child goes black as it struggles for breath Convulsive mov may occur and occasionally consciousness is lost The nature attack may lead to confusion with the convulsive seizures of e and equally the epileptic fit in childhood is often mistaken for attack The combined effects of anoxaemia and polycythaem responsible for angina pectoris in the case of Blackford (1930)

Polycythaemia has its attendant disadvantages and dangers is a tendency to thrombosis in cerebral vessels and in other ve the body Cerebral thrombotic attacks are accompanied by con and often a hemiplegia which may be permanent (Perry 1942 and Lipscomb 1942) A further effect of increased viscosity blood is peripheral stasis and capillary dilatation and hence in deoxygenation Packing of the alveolar capillaries with blo

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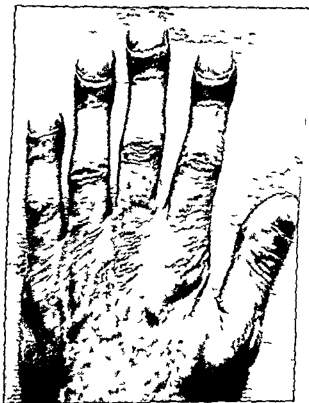


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contemplated as a means of relief of the more urgent symptoms The full details of diagnosis are discussed under the appropriate lesions and only principles are discussed here

The X ray screening which can be conducted in the out patient department is of the greatest importance and attention should be directed particularly to the pulmonary artery and conus Absence of the normal pulmonary conus a concavity at the site of the pulmonary arc a small pulmonary artery and diminished or absent hilar pulsation and abnormally clear lung fields all suggest that the fundamental difficulty is a diminished pulmonary circulation The X ray photograph in antero-posterior and left anterior oblique views may be very helpful The peppered vascular shadows and ischaemic lung fields are quite distinctive of an inadequate pulmonary circulation Conversely a large pulmonary artery actively pulsating a full pulmonary conus and evidence of congestion in the lung fields all imply an adequate or increased pulmonary circulation and that the cause of cyanosis must be sought elsewhere

The fuller investigation of a case is best carried out with the patient admitted to hospital It is well to have a definite plan of events spread over a few days The oxygen saturation of the blood is estimated from a sample of blood withdrawn from the femoral artery Even in the presence of the anatomical changes of the tetralogy an oxygen saturation of 75 per cent suggests the inadvisability of an operation which will only raise saturation a little over 80 per cent The effects of prolonged inhalation of oxygen may also be studied By this means it may be possible to greatly modify or even cause the disappearance of cyanosis but the level of oxygen saturation so induced never reaches a normal figure If there is a shunt from the venous side of the heart as in those anomalies with a dextroposed aorta the amount of oxygen unsaturation as influenced by the volume of the venous shunt can never be rendered normal by oxygen administration It is thus possible by comparison of figures obtained before and after oxygen administration to assess the relative importance of the pulmonary factor (influenced by oxygen inhalation) and the venous shunt not so influenced

The circulation may also be measured by various methods in order to ascertain or confirm the presence of a venous shunt The circulation time tends to be reduced in anomalies with a dextroposed aorta

Lastly there are those methods which involve intracardiac catheterisation These should properly be reserved for the more complex cases which defy the usual methods of diagnosis Similar considerations apply to angiocardiography a difficult technical performance and not to be lightly undertaken because it carries some risks Angiocardiography has made its most important contribution to cardiology in the study of the cyanotic case It demonstrates the permeability of the pulmonary tract and the presence of pulmonary arteries

PULMONARY STENOSIS AND ATRESIA

The cases in this group naturally fall into four main types dependent upon the presence or absence of a defect of the interventricular septum Cases with pulmonary stenosis or atresia with a defect of the inter ventricular septum are usually considered together as the tetralogy of Fallot With the exception of a few cases of valvular pulmonary stenosis with a closed ventricular septum the group as a whole is the result of abnormality in the involution of the bulbus cordis This structure plays a prominent role in the early development of the heart and complex abnormalities result when its involution is disturbed The bulbus cordis is a dominant and permanent structure in the hearts of the lower vertebrates It exists as a separate chamber in the teleost and elasmobranch fishes and in some reptiles In the mammalia the bulbus is incorporated into the right ventricle where it forms the conus or infundibulum It serves to protect the pulmonary arterioles and capil laries against rises in blood pressure promoted by exercise and increased activity of the heart (Keith 1909) It is stated that the musculature of the infundibulum is activated later than that of the sinus of the right ventricle

PATHOGENESIS The work of Keith (1909) has largely elucidated the problems connected with the fate of the bulbus cordis in the human heart It is a transitory structure which disappears in the left ventricle and is mainly incorporated into the right ventricle to form the in fundibulum Its persistence in the left ventricle leads to subaortic stenosis The bulbus also contributes to the development of the upper part of the septum (bulbar septum) and to the aortic and pulmonary valves Abnormalities in its involution are also closely connected with the problems of transposition of the great vessels Upon the degree of expansion or incorporation of the bulbus cordis depend a gradation of cardiac defects ranging from division of the right ventricle into two chambers down to complete pulmonary atresia To quote Keith a very large number of deformed hearts are the result of an arrest in that process which ends in incorporation of the bulbus in the right ventricle

To this process must be added the effects of torsion As the animal scale is ascended there is a transition from the purely gill breathing types to the lung breathing types with the result that the pulmonary portion of the cardiac tube becomes at least as important as the

systemic part Two systems thus develop side by side separated by septa In the early stages of development when the septa are beginning to form this process of torsion occurs Examination of the normal heart will show that if the aorta and pulmonary artery are to assume their correct positions relative to their respective ventricles a torsion or rotation of about 180 is necessary Evidence of this rotation is found in the clockwise course of the pulmonary artery about the aorta Where no rotation has occurred or where there has been detorsion there is a change in this relationship and varying degrees of transposition of the vessels results The most extreme degree of such transposition is found in transposition of the vessels with a closed ventricular septum where the aorta arises from the right ventricle and the pulmonary artery from the left The least degree of torsion with the slightest dextroposition of the aorta may be found in congenital aneurysm of the interventricular septum This conception of torsion is of great importance because it serves as a basis of any explanation of certain severe cardiac defects

Spitzer s (1923) theory seeks to establish a phylogenetic relationship between the human and the reptilian heart In the normal human heart he recognises the site of the closed reptilian right aorta by a niche in the right ventricle situated between the crista supraventricularis and the infundibular cusp of the tricuspid valve At an early stage the reptilian right aorta is closed by the clockwise rotation of the bulboventricular end of the primitive cardiac tube If for any reason this does not occur there is an apparent detorsion in a counter clockwise direction which brings the parts out of position and results in reopening of the reptilian right aorta with closure of the left If the degree of detorsion is small these two aortae may fuse and give rise to a single larger than normal dextroposed aorta Such a theory fits in with the observed facts In the tetralogy of Fallot there is a large dextroposed aorta and with singular frequency a bicuspid pulmonary valve homologous with that found in the reptile There has been some criticism of Spitzer s theory notably by Lev and Saphir (1927) who admit that it is valid anatomically but disagree with the mode of its accomplishment (Chapter XVII) They believe that an abnormality in the bulboauricular spur area prevents absorption of the bulbus into the left ventricle thereby defaulting the normal clockwise rotation about this point As a consequence the unfixed lower end of the bulbus rotates in a counter clockwise direction and this leads to a more or less parallel position of the aorta and pulmonary artery instead of the habitual spiral twisting Further there is imperfect union or no union at all between the lower end of the aorticopulmonary septum and the ventricular septum resulting in ventricular septal defect

Delayed involution of the bulbus cordis results in conus or infra valvular stenosis of the pulmonary artery The foramen between the

conus and the sinus of the right ventricle is the lower bulbar orifice or ostium infundibuli of Keith The crista supraventricularis marks the line of division which separates the bulbus cordis from the common ventricle in the fourth and fifth weeks and in cases where there has been persistence of the bulbus cordis the crista is a prominent structure in contrast to its usual insignificance in the normal right ventricle There is often a small interventricular septal defect opening into the sinus of the right ventricle but it is not uncommon for this to be absent Some degree of dextroposition may also be present



FIG 62 Conus stenosis of the pulmonary artery (After Laubry)

In cases where there is an arrest of the developmental expansion of the infundibulum the infundibulum is foreshortened and exists as a small fusiform cavity with thickened walls In about half the cases there is also fusion of the pulmonary cusps The lining of the conus is of thickened endocardium of a peculiar ground glass appearance attributed by Keith to embryonic fibrocellular tissue An interventricular septal defect may be present

Where the arrest of expansion has been complete there is pulmonary atresia The orifice of the pulmonary artery is represented by a mere depression or a mass of scar tissue The first part of the pulmonary artery may be an impermeable fibrous cord This expands into the two main branches of the pulmonary artery which are often thin walled and hypoplastic The left ventricle is small and aplastic and the auricles dilated and hypertrophied A defect of the interauricular septum is frequently present The dextroposed aorta arises wholly or partially from the right ventricle The ductus arteriosus may be widely patent and there may be anomalous vessels or hypertrophied bronchial arteries assuring a blood supply to the lungs Such cases come under the general category of the tetralogy of Fallot In cases where there is no defect of the interventricular septum the right ventricle is aplastic

the aorta larger than normal the ductus arteriosus patent and again there is a collateral pulmonary circulation developed from bronchial and other arteries

The purely valvular type of pulmonary stenosis has been stated to be the result of a foetal endocarditis involving the pulmonary cusps after the septa have closed This leads to their fusion and deformity which may be so severe as to amount to atresia Abbott (1927) states that practically all cases of valvular pulmonary stenosis with closed

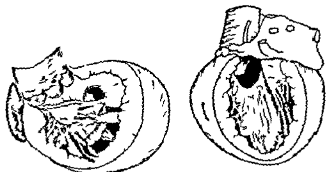


Fig 63 Pulmonary atresia in a child aged 2½ years The aorta arises wholly from the right ventricle which is hypertrophied The pulmonary artery is reduced to a fibrous cord The circulation was carried on through hypertrophied bronchial arteries

septa arise by this means Gross (1941) suggests that the accumulated evidence throws doubt upon the existence of a foetal endocarditis of as yet unknown etiology In his view it is probable that abnormalities of this valvular type arise from arrest of development of or closure of arteries supplying the valve resulting in infarcts and subsequent fibrosis Microscopical examination of valves reveals only moderate fibrosis and none of the stigmata of the inflammatory diseases The fusion of the pulmonary cusps into a cupola of symmetrical proportions observed in some cases of valvular stenosis is quite unlike an inflammatory scarring process Further the arrangement of the cusps is altered so that in pure pulmonary stenosis there are one anterior and two posterior cusps The presence of other anomalies either cardiac or somatic is in favour of a developmental origin Keith (1909) has drawn attention to the frequency with which valvular and infundibular stenosis are associated

Pulmonary atresia may be in a few cases the result of inflammatory myocardial changes leading to fibrosis in the conus and occurring after the septa have closed In some cases syphilis has been suggested as the cause of myocarditis in this situation but the evidence is not at all convincing

PULMONARY STENOSIS WITH CLOSED VENTRICULAR SEPTUM

Pulmonary stenosis with a closed *ventricular septum is statistically* rarer than the tetralogy of Fallot Clinical experience does not altogether support this view of rarity In Abbott s 1 000 cases there were twenty five with an intact ventricular septum and sixteen of these had a patent foramen ovale Currens *et al* (1945) have added a further eleven cases five with a patent foramen *ovale To these may be added the* single cases of Blackford and Parker (1941) Garrison and Feldt (1942)

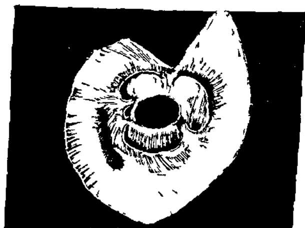


FIG 64 Isolated pulmonary stenosis Pulmonary valve viewed from above The cusps are fused into a diaphragm

Kinney and White (1945) Freed and Budnitz (1946) and Auerbach and Harper (1947)

CLASSIFICATION AND TYPES There are several varieties of pulmonary stenosis and these are classified according to the site of the lesion

(1) *Valvular Stenosis* In this condition the cusps are often abnormally arranged there being one anterior and two posterior (fig 64) The pulmonary cusps are *fused and thickened so as to form a diaphragm* with a central opening of varying dimensions This diaphragm may resemble a funnel or dome projecting into the pulmonary artery with an aperture at its peak a very good example being figured by Peacock (1866) Above the stenosis the pulmonary artery is generally dilated *The right ventricle and conus are hypertrophied and often dilated In* a number of cases the brunt of the stenosis falls upon the pulmonary annulus which is contracted and fibrosed *Accompanying this there* may be scarring of the pulmonary cusps The foramen ovale is frequently patent either primarily or in response to raised pressure *in the right side of the heart*

These cases are stated to be rare but this opinion probably requires revision Cardiac catheterisation and angiocardiography will probably result in the more frequent recognition of this anomaly

(2) *Conus Stenosis (Infravalvular Stenosis Stenosis at the Lower Bulbar Orifice)* In this lesion first differentiated by Keith (1909) but known to Peacock (1866) the conus of the right ventricle is dilated and forms a separate chamber or cavity communicating above with the dilated but otherwise normal pulmonary artery Below it communicates with the sinus of the right ventricle by a lower bulbar orifice or ostium infundibuli This differentiation into two cavities is effected by a band of muscular tissue encircling the wall of the ventricle and is in fact the crista supraventricularis It represents persistence of the bulbus Various degrees of conus stenosis may exist In many cases an interventricular septal defect is associated and opens beneath the ostium infundibuli into the sinus of the right ventricle The ostium is lined with fibrous tissue and together with the wall of the cavity may become the seat of infective endocarditis Examples of conus stenosis are figured by Keith (1909) and Laubry (1930) (fig 62) and Konwaler (1944) Excellent examples with infective endocarditis are those of Bedford and Brown (1937) (fig 65) and Dryerre and Walmsley (1939) Eakin and Abbott (1933) have reported two cases in the first of which there was no defect of the interventricular septum but aneurysmal bulging of the upper part of the septum and slight dextroposition of the aorta In their second case again devoid of an interventricular septal defect infective endocarditis involved the lower bulbar orifice and pulmonary cusps In a rarer type of conus stenosis the infundibulum is atrophied and the conus consists of a short narrow passage

In both valvular and conus stenosis the pulmonary artery may be dilated distal to the stenosis the dilatation extending as far as the bifurcation or beyond The reason for such dilatation is not clear but it may depend upon a hypoplasic condition of the artery wall Ligature of the pulmonary artery in animals is stated to lead to dilatation distal to the ligature and the microscopic changes of diminution of the media In some cases the dilatation appears to be dynamic and only recognisable by radiological examination during life It is absent at autopsy

An unusual type of pulmonary stenosis has been reported by Allen (1941) In a case with a defect of the interventricular septum the base of the right aortic cusp was fused to the anterior pulmonary cusp the aortic cusp being eventrated through the septal defect into the right ventricle to cause a pulmonary stenosis This peculiar structure was the seat of a bacterial endocarditis

Clinical Picture Cyanosis is usually only slight and late in onset often not appearing until early adult life and in some cases as late as the fourth decade It depends upon permeability of the foramen ovale and upon the degree of pulmonary obstruction If this latter is of a

severe nature cyanosis may then be early and there may be little on clinical grounds to distinguish the case from the more common tetralogy of Fallot In a few cases *cyanosis may never appear despite obvious* physical signs of pulmonary stenosis Once established it is slowly progressive and may become extreme with failure The cyanosis results from several causes the more important being a diminished blood supply to the lungs a right to left shunt through an auricular *septal defect and pulmonary* arteriolar disease and changes in the



Fig 65 Stenosis of conus arteriosus of right ventricle below pulmonary cusps with *malignant endocarditis at site of stenosis* Pulmonary artery much dilated beyond stenosis aorta slightly constricted at isthmus

lungs interfering with oxygenation Clubbing of the fingers is proportional to the cyanosis Exertion which increases oxygen consumption and emphasises cyanosis may be the basis of a simple clinical test in a doubtful case

The principal symptoms are dyspnoea on exertion and paroxysmal attacks of dyspnoea accompanied by cyanosis may occur Such attacks may develop years before the appearance of permanent clinical

cyanosis Epistaxis is not uncommon Incapacity is never so great as in pulmonary stenosis with a ventricular septal defect (Fallot) except in the rather rare cases of severe valvular stenosis amounting to atresia with a grossly diminished pulmonary blood flow and a closed ventricular septum When failure is present cyanosis becomes marked and dyspnoea a prominent symptom Failure tends to be progressive and not very amenable to treatment Squatting is only observed in the very severe cases

The physical signs are a harsh systolic murmur and accompanying systolic thrill in the second left interspace close to the sternum The murmur is generally harsher and louder than that heard in the tetralogy and is conducted towards the left clavicle and often into the left side of the neck Occasionally both thrill and murmur may be maximal in the second right interspace and in a non cyanotic case be confused with aortic stenosis Often the systolic murmur is audible over the entire praecordium and may be heard at the angle of the left scapula Its loudness is due to the nearness of the vessel to the stethoscope The pulmonary second sound is rarely normal but is reduced in intensity or frequently absent When confronted with the anatomical specimen it is often difficult to understand that the pulmonary second sound has been heard at all Perhaps in some cases the aortic second sound is transmitted to the pulmonary area and reinforces a diminished pulmonary second sound There may be dullness in the upper left intercostal spaces corresponding to a dilated pulmonary artery In conus stenosis the murmur is placed lower and is systolic in time Its maximum intensity is in the fourth right space close to the sternum (Keith 1909)

RADIOLOGY The heart is normal in size or only slightly hypertrophied (fig 66) When failure is present the heart is grossly enlarged Hypertrophy involves the right ventricle and the apex of the heart may be blunt and turned up giving a coeur en sabot silhouette The right border of the heart may be formed by the hypertrophied right ventricle which pushes the right auricle backwards so that only its appendage contributes to the upper part of the silhouette This may be recognized by the synchronous pulsation of both borders of the heart The pulmonary artery is most often dilated and may be a prominent feature of the left cardiac border Dilatation and hypertrophy of the conus may produce similar changes on the left border (Usomoto 1925)

ELECTROCARDIOGRAM The electrocardiogram usually shows a right axis deviation often to the extent of that seen in the tetralogy of Fallot In the case of Wood (1942) where pulmonary stenosis was combined with a large auricular septal defect there was a left axis the only example so far reported A normal axis has been observed by Currens Kinney and White (1945) and others so that it appears that a normal axis does not entirely preclude a diagnosis of isolated pulmonary

stenosis A high peaked P wave is common in lead 2 and the T wave in leads 2 and 3 may be inverted Conduction abnormalities are not uncommon and right bundle branch block has been described by Blackford and Parker (1941) and others Intraventricular block occurred in the case of Auerbach and Harper (1947)

COURSE AND PROGNOSIS This naturally depends upon the severity of the stenotic lesion and its effects upon the pulmonary blood flow In slight or moderate degrees of stenosis the patient is often in good



FIG 66 Pulmonary stenosis with closed ventricular septum Female aged 28 Three pregnancies well tolerated and offspring normal

health and able to engage in ordinary employment and pleasure until the second or third decades Cyanosis and dyspnoea may then become troublesome together with a greatly reduced physical capacity Disability is only severe in those cases where the stenosis is of high degree and the clinical picture may then resemble that of the tetralogy of Fallot Death results from infective endocarditis cardiac failure or pulmonary tuberculosis Personal cases have died of phthisis (2) and malignant hypertension In Abbott s (1931) series the average age at death was 20 6 years with an extreme of 57 years

This abnormality might well be one of the conditions for which surgery may be beneficial in prolonging life An operative attempt to sever a constricted valve was made by Doyen (1913) but failed owing to the presence of subvalvular stenosis The Blalock Taussig procedure should be considered as applicable to severe and progressive cases

with much disability Valvulotomy has been successfully performed by Holmes Sellors (1948) and Brock (1948)

DIAGNOSIS The principal physical sign is a harsh systolic murmur with accompanying systolic thrill in the second left interspace close to the sternum Its loudness the thrill and the conduction of the murmur to the left clavicle and the left carotid artery help in its differentiation from an innocent pulmonary systolic murmur The X ray picture shows a dilated pulmonary conus and artery The dilatation does not always extend to *the branches and if it does it is* never to the degree of that found in the Eisenmenger complex or auricular septal defect The usually prominent pulmonary border is in contrast to the concave pulmonary arc of the tetralogy The auricular septal defect is further distinguished by its large heart and enlarged pulmonary branches Angiocardiography *may be successful* in demonstrating a valvular or conus stenosis but it is not always reliable A permeable pulmonary trunk must be present if valvulotomy is to be performed The electro cardiogram almost always displays a right axis and conduction defects are not nearly so common as in the auricular septal defect In a doubtful case *a normal circulation* time will favour a closed ventricular septum although the method is not infallible Ten to even teen seconds from arm to tongue may be taken as a normal figure this tending to be shortened in conditions with a right to left shunt such as the tetralogy The most positive method of confirmation of isolated pulmonary *stenosis is by catheterization* Pressure will be found to be lower in the pulmonary artery than in the right ventricle

Gouley (1937) has shown that stenosis of the pulmonary artery may be among the effects of an adhesive pericarditis and has confirmed this by autopsy control Thickened adherent pericardium surrounds the pulmonary artery leading to a notable *diminution* in its calibre The recognition of these cases depends upon a history of rheumatism evidence of rheumatic valvular disease and signs of an adherent *pericardium* A loud banging pulmonary second sound is commonly present Radiological examination shows an enlarged heart and no dilation of the pulmonary artery although the *conus may be expanded* Ayerza s disease has occasionally caused difficulty but it is recognized by its history serology and the very marked cyanosis

PULMONARY STENOSIS OR ATRESIA WITH DEFECT OF THE INTERVENTRICULAR SEPTUM THE TETRALOGY OF FALLOT

This is a well defined entity in congenital heart disease of the cyanotic type Essentially it consists of stenosis hypoplasia or atresia of the pulmonary artery of developmental type together with an interventricular septal defect dextroposition of the aorta and hyper *trophy of the right* ventricle

HISTORICAL This combination of lesions was first reported figured by Sandifort in 1777 Bennett s (1946) admirable transl of this work should be consulted Sandifort s figures clearly sho aorta arising from both ventricles and the clinical observations com very favourably with present day knowledge Farre (1814) was pressed by the frequency of pulmonary stenosis interventricular s defect and overriding aorta Corvisart (1815) describes a case wi right aortic arch The condition was first diagnosed during life by H (1839) Much information relative to the earlier cases may be foun the pages of Peacock (1866) Peacock was well familiar with frequency of this combination of defects and figures a case wi right aortic arch He considered the usual interventricular septal de to be the homologue of the foramen between the aortic ventricle the turtle He also preceded Spitzer in his belief that the sinus infundibular parts of the human right ventricle corresponded to right systemic and pulmonary ventricles of the turtle Fallot (18 made no new contribution to the anatomy of the abnormality emphasized the frequency with which this combination of def occurred and the name tetralogy arose from the four constar present anatomical defects He made it clear that the lesion might recognised on clinical grounds and stated Until now clinicians h considered the precise diagnosis of anatomical lesions of the morb caeruleus an almost insurmountable difficulty On the contra we see from our observations that cyanosis especially in the adult the result of a small number of cardiac malformations well determined Perhaps here we have some slight justification for the eponymo designation of the tetralogy of Fallot Fallot s analysis of report congenital cyanotic cases and his own showed that 74 per cent we examples of the tetralogy He asserted that a physician confronted wi a congenital cyanotic case might justifiably diagnose the tetralogy would be idle to mention all the cases reported since that time b mention may be made of the fundamental work of Abbott (1927) I recent times the operation devised by Blalock and Taussig (1945) wi its possibility of surgical relief of cyanosis has aroused great interes and made accurate diagnosis of paramount importance

ANATOMY While it is true to say that the essential features of th tetralogy are those defined above these features may vary within wid limits from case to case The variables are the degree of pulmonar stenosis or atresia the situation of the aorta vis a vis the defect of th interventricular septum and alterations in the course of the aorta

The situation of the interventricular septal defect may also vary and it may open most often into the sinus of the right ventricle and occasionally into the conus of the right ventricle In the form r case the defect is beneath the non coronary and right coronary cusps and

is more anterior and beneath the right coronary cusp with the chordae of the infundibular cusp of the tricuspid valve attached to its free margin The defect itself in either case is variable in size crescentic in shape and has a smooth fleshy margin its concavity being directed upwards (fig 67) This contrasts with the lesion found in the maladie de Roger where the defect is circular and ringed with fibrous tissue The crista supraventricularis is usually a striking feature and is a hypertrophied mass of tissue intervening between the conus and septal defect

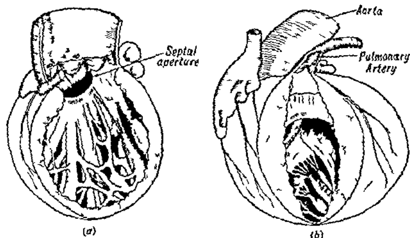


FIG 67 Fallot s tetralogy (a) Left ventricle opened showing defect of septum (b) Right ventricle opened showing hypoplasia of pulmonary artery and large aorta

and helps to divert blood from the right to the left ventricle Most often the conus of the right ventricle is small and hypoplastic with a slit like cavity In those cases where the ventricular septal defect opens into the conus the cavity is large and thick walled so that this may cause an alteration in the radiograph and is thus a feature of importance The aorta is dextroposed and Abbott (1923) has pointed out that in cases where there is a dextroposition of the aorta the membranous septum lies between the basis of the noncoronary and left coronary cusps The non coronary cusp is thus in front of the membranous septum instead of in its normal position behind In spite of its apparent change in position it still bears its normal relationship to the right auricle and ventricle A needle passed through the anterior portion pierces the septal cusp of the tricuspid valve and passed through the posterior portion it reaches the right auricle above the attachment of the septal cusp This maintenance of a normal relationship shows that the change

of position is entirely of the aorta In dextroposition the aortic cusps are rotated in a clockwise direction

ASSOCIATED ABNORMALITIES An important and striking associated abnormality is a right aortic arch which occurs in at least 20 per cent of cases Corvisart (1815) was the first to describe *this association with* congenital heart disease of cyanotic type Peacock (1856) figured and described this anomaly Of recent years additional examples have been presented by Assmann (1924) Bedford (1929) and Lian and Marchal



FIG 68 The tetralogy of Fallot in a woman aged 23 who died of pulmonary arteritis The interventricular septal defect opened into the conus of the pulmonary artery The pulmonary artery was bicuspid and vegetations are seen distal to the pulmonary cusps

(1937) Bedford and Parkinson (1936) emphasized its frequency in reporting four additional cases The surgical experience of Taussig and Blalock stresses its frequency and it appears that between 20 and 25 per cent of cases of the tetralogy have a right aortic arch Its presence or absence *determines the site of the incision at operation and it is there*fore important that it should be recognized

The foramen ovale may or may not be patent The earlier observers attributed cyanosis to patency of the foramen despite the refutation of Fallot Although an interauricular septal defect may appear to prolong *life in such conditions as the* Lutembacher syndrome Abbott s (1931) statistics indicate that an open or closed foramen ovale has no material influence on the duration of life

Exceptionally inflammatory changes may be found in the pulmonary cusps which become thickened and deformed In Halpert and Tennant s (1934) case there was a healed pulmonary valvulitis and a diffuse myocarditis of the right ventricle The case of Campbell and Coulthard (1924) has probably been wrongly attributed to an inflammatory process

In regard to the pulmonary artery The degree of stenosis is highly variable and may be from a slight narrowing down to complete atresia The pulmonary artery is thin walled and may have the appearance of a vein It may be reduced to a fibrous cord (fig 63) and in these circumstances a pulmonary blood flow is maintained by a patent ductus arteriosus or by a collateral circulation developed through bronchial and other arteries or both The site of stenosis may be valvular or sub valvular The practical issue is that in most cases the infundibulum is narrowed as well as the pulmonary vessel itself The infundibulum becomes a narrow canal (retresissement canaliculaire of Laubry) Occasionally the main site of stenosis is at the lower bulbar orifice The pulmonary circulation is maintained at a very limited level through these narrow but often permeable routes In cases of extreme stenosis or complete atresia the ductus arteriosus remains patent to assure a pulmonary circulation This is a point of practical importance because the spon taneous closure of the ductus at any age may lead to the death of the subject Patency of the ductus can often be recognized clinically and implies pulmonary atresia Such a ductus should never be ligatured

Collateral Circulation to the Lungs

The importance of a collateral circulation to the lungs in severe cases of the tetralogy and in pulmonary atresia is now well recognized Christeller (1916) stressed the role not only of the bronchial arteries but also of collaterals rising from the mediastinal oesophageal in ternal mammary pericardial and other arteries such as the subclavian and even the coronary arteries Collaterals most commonly arise from the bronchial and mediastinal arteries These vessels are enlarged and may be sufficiently large as to distort the barium filled oesophagus or cause notching of the under surface of a rib

The existence of bronchial arteries has been known from the earliest times Ruysch (1696) pointed out that they were separate from but unastomosed with the pulmonary artery system and his illustrations are possibly the first that were ever published of these structures It is clear that they were known before his time even if their origin and distribution had not been accurately delineated In severe pulmonary stenosis and atresia the bronchial arteries are greatly hypertrophied and the branches may be as large as the parent trunk Pre capillary anastomosis between bronchial and pulmonary arteries has been observed near the hilum by Hales and Liebow (1948) and has been

experimentally produced in dogs by ligation of the pulmonary artery as in the work of Mathes Holman and Reichert (1932) Similar anastomoses have also been observed in situations more remote from the *hilum* in bronchiectasis It must be realised that the bronchial arteries are under a high systemic pressure and this combined with the largeness of their branches allows a considerable volume of blood to pass through an anastomotic channel Severe pulmonary stenosis furnishes a stimulus to the development of a bronchial artery circulation and the frequently observed thrombosis in the smaller branches of the pulmonary artery embarrassing an already embarrassed pulmonary circulation acts in the same way

The general anatomy of the bronchial arteries and their functions has been reviewed by Cauldwell *et al* (1948) These vessels most often arise either independently or in multiples from a common stem Usually there are two right and one left arteries but there may be one left and one right or two arteries or more to each side The right bronchial arteries usually arise in common with an aortic intercostal artery usually the first right aortic intercostal occasionally the second or third The left bronchial arteries generally spring from the anterior aspect of the aorta or from the concavity of the arch and rarely in association with an intercostal artery Sometimes right and left vessels and particularly the inferior bronchial vessels may arise from a common trunk but this is unusual Bronchial arteries may also arise from the subclavian vessels Their distribution is to the bronchi the mid portion of the oesophagus pericardium and they anastomose with neighbouring vessels

The efficacy of a collateral circulation is well demonstrated by the cases of Finlay (1930) East and Barnard (1938) and Volini and Flaxman (1938) In rare cases mediastinal and oesophageal vessels may also participate in this collateral circulation In Christeller s (1917) well known case a large upper oesophageal artery arose just below the third left intercostal artery and supplied both lungs Cases where there is an adequate collateral circulation may survive into middle adult life or later Two cases of East and Barnard survived to 30 and 20 years Vose (1856) case to 37 years Herzog s (1919) case was remarkable in that the collateral circulation was provided by union of a branch of the right inferior thyroid with the internal mammary and bronchial arteries to form a rich vascular plexus

CIRCULATION The general effects of pulmonary stenosis on the circulation are discussed elsewhere In pulmonary atresia the presence of a defect of the interventricular septum tends to prolong life by affording relief to the right side of the heart always provided that there is some accessory pulmonary circulation provided by a ductus arteriosus or bronchial arteries or both In the tetralogy there may be a definite handicap because the size of the defect and the position of

the aorta in relation to the defect straddling both ventricles facilitates a large venous arterial shunt This shunt is proportional to the degree of obstruction in the pulmonary artery and the output of the heart

Physiological and Pathological Results of the Anatomical Defects

Cyanosis exists from infancy and increases with age It may not be very evident until several months after birth Cyanosis in the tetralogy is due to the combined effects of a venous arterial shunt into a dextroposed aorta and most important of all as emphasized by the work of Taussig and Blalock (1925) upon the amount of blood reaching the lungs for oxygenation It is evident that both these factors will be influenced by the degree of pulmonary stenosis and marked from birth in atresia Secondary factors are increased deoxygenation in the capillaries increased viscosity of the blood and polycythaemia of itself leads to changes in the lungs that inhibit effective contact of capillary blood with oxygen in the alveoli

The combined effects of shunt and pulmonary obstruction lead to right ventricular hypertrophy The circulation time as measured by decholin is usually reduced because a certain amount of the substance escapes the pulmonary circulation and is shunted into the dextroposed aorta Normally about 10-12 seconds times as brief as four seconds have been noted by Brumlik (1937) and McGuire and Goldman (1937) Such reduced times will not be so marked when stenosis is of lesser degree and it must not be forgotten that in the absence of pulmonary stenosis as in the Eisenmenger complex a similar slight shortening of circulation time may be present Modern methods of intracardiac catheterization allow precise investigation of the dynamics of the heart in these cases and permit a precise estimation of the volume of a venous arterial shunt and of the total volume of blood reaching the lungs for aeration The reader is referred for details of this highly technical work to the reports of Bing *et al* (1947)

Classification of Cases

In the light of the above considerations it is possible to divide cases of the tetralogy on a clinical basis into the following categories

(1) Cases with pulmonary stenosis allowing a sufficient pulmonary blood supply to maintain life The ductus is closed
(2) Cases with functional or anatomical pulmonary atresia The ductus remains patent and a collateral circulation is maintained through bronchial and other vessels
(3) Cases with extreme dextroposition of the aorta so that the aorta arises almost wholly from the right ventricle

(4) Cases with a right aortic arch but with the same general characters as the above groups

(5) Bizarre forms accompanied by transposition of the vessels or by dextrocardia

CLINICAL PICTURE Cyanosis appears early and once established is progressive It varies from case to case and may be present from birth in about one third of the cases or makes its appearance in later infancy or early childhood when the child becomes more active Cyanosis may appear for the first time during some intercurrent illness or it may be detected during a routine examination often undertaken because the child has failed to develop at a normal rate In a personal case of the tetralogy established cyanosis was never present during life and the presence of the typical lesion was established at autopsy at the age of 29 Similar cases have been reported by Galliard and Cawadias (1908) and by Fleury (1937) Such cases are very exceptional In general it may be said that the transition from the vegetative state of infancy to the fuller activities of early childhood may make cyanosis appear for the first time or become obvious to the observer Cyanosis naturally depends upon the severity of the lesion and is intensified by exertion or emotion It is most marked in the mucous membranes and extremities It may be paroxysmal without apparent cause A sudden increase in cyanosis and deterioration of the patient may mark the spontaneous closure of a hitherto patent ductus arteriosus As cyanosis develops clubbing appears and is proportional to the degree of cyanosis It is rare to find clubbing before the age of two but once established it may rapidly become extreme If anaemia is present clubbing may be absent or late in appearance

Symptoms are numerous and are mainly referable to the cyanosis Many cases are without dyspnoea at rest but the least exertion may provoke extreme breathlessness with intensification of cyanosis Dyspnoea appears to be proportional to the severity of the anatomical defect and its degree may mould the whole of the patient s life Some cases can play but never with the vigour and abandon exhibited by their fellows In others mere change of posture provokes intense dyspnoea and for them life is a case of existing rather than living Helen Taussig observes that the child with the tetralogy when exhausted by exertion assumes a squatting position and believes that such a posture is an important diagnostic point Exertion may also precipitate attacks of dyspnoea of suffocating intensity and similar attacks may arise unexpectedly without apparent cause *The essential* features of what the patient may term an attack are increasing dyspnoea terminating in asphyxia when the patient goes black and loses consciousness Convulsive movements of the limbs occur and the attack may have a fatal termination Some observers have noted a similarity to Stokes Adams seizures and Nobecourt (1925) showed that

complete heart block was present with a pulse rate of 64 in a child of 10 months during an attack Relatively few of these attacks have a similar etiology Indubitably true epilepsy occurs in some of the cases Abbott (1927) considers that these attacks are due to the combined effects of anoxaemia and polycythaemia which might cause small areas of capillary thrombosis in the brain Analogous changes may be found in the brain in Vaquez s disease There are also the head aches and vertiginous attacks followed by convulsive movements and leaving residual hemiplegias which herald the onset of cerebral throm botic incidents or of the embolic accidents of infective endocarditis Dextroposition of the aorta favours the occurrence of paradoxical embolism and many cases have presented a closing clinical picture of cerebral abscess

Polycythaemia as an effort to increase the oxygen-carrying capacity of the blood soon develops in the severe case In the tetralogy the red cell count may average eight or nine million The count may rise as high as twelve million and a high rising count corresponds with deterioration in the patient s condition and may be an important indication for early operation In the present state of knowledge it is unwise to attach too much significance to an isolated count Poly cythaemia leads to congestive changes in the viscera a tendency that is reflected in the occurrence of epistaxis haemoptysis and even haematemesis Haemorrhages may be temporarily beneficial but are a sign of the gravity of the case The blood pressure tends to be low Orthostatic albuminuria is frequent In Blackford s (1930) case there was angina of effort at the age of 14 This was considered to be due to anoxaemia Angina similarly occurs in polycythaemia vera (Kahn 1926)

Squatting which has been discussed on a previous page occurs in about 75 per cent of the younger patients and to some extent reflects the degree of anoxaemia present In older subjects the habit may become camouflaged by a pretence of tieing a hoe lace No explana tion is as yet forthcoming as to the mechanism of relief by this manoeuvre

Enlargement of the heart is exceptional and when present involves only the right ventricle A systolic thrill may be present but not so fre quently as in isolated pulmonary stenosis If present it is maximum in the pulmonary area but it may be felt diffusely over the praecordium Rarely a diastolic shock is palpable at the base A continuous thrill may be present if there is a patent ductus arteriosus but is an excep tional observation The auscultatory findings are not pathognomonic A systolic murmur is present in at least 90 per cent of cases and is most pronounced in the second and third left interspaces It may be audible over the entire praecordium and is transmitted towards the left clavicle and into the vessels of the neck It may also be heard at the

angle of the left scapula Conduction of the murmur into the neck ha been urged by Laubry and Pezzi (1921) and others as a capital poin in the diagnosis of the tetralogy and as a point of distinction from the murmur of isolated pulmonary stenosis but this idea is not tenable Conduction into the neck is due to dextroposition of the aorta and should be compared with the relatively rare transmission of the murmur in the isolated ventricular septal defect Rarely the murmur is entirely absent this being likely in pulmonary atresia or where a large septal

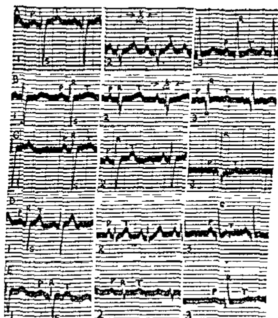


Fig 69 The electrocardiogram in five cases of the tetralogy of Fallot

defect is unfavourable for the production of any murmur The contrast between the murmurs of the isolated components of the tetralogy and their combination is striking because the murmurs of the combined defects are much less prominent In the tetralogy hypoplasia of the pulmonary artery anatomically consists of a long tube of narrow calibre rather than the sudden constriction and then widening of a pure pulmonary stenosis which latter is so favourable for the produc tion of a bruit Bard (1921) was of the opinion that the large septal defect was the principal factor causing the diminution in the intensity of the murmur A large defect allows blood to pass along a new route with lowering of pressure in the right ventricle and hence a lower

re of blood entering the hypoplastic pulmonary artery A dia murmur may occasionally be heard and is due either to coinci ortic incompetence from a large aorta or to a patent ductus A uous murmur betokens a patent ductus arteriosus

s often stated that the pulmonary second sound is reduced or in the tetralogy This is not the case for the second sound is lly audible and is at least as loud as the aortic second sound and ny cases louder If the pulmonary second sound is reduplicated

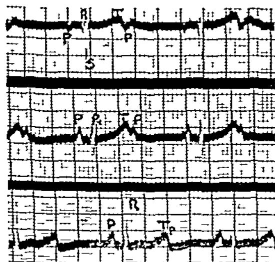


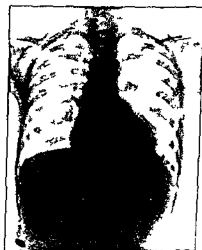
FIG 70 Tetralogy of Fallot Female aged 6 years Marked right axis with 2 to 1 block

ggests the presence of two functioning vessels A loud second sound he right of the sternum may occur when there is considerable roposition or a right aortic arch

LECTROCARDIOGRAM Right axis deviation of moderate or marked ee is found in every case of the tetralogy (fig 69) unless complicated dextrocardia or gross conduction defect Thus the finding of mal or left axis deviation in a cyanotic case in the absence of these plications should exclude a diagnosis of the tetralogy While the t extreme degrees of right axis deviation are found in the tetralogy ventricular complexes are not invariably abnormally tall The aves *are often high and* pointed especially in lead 2 *Conduction* ects occur as in other cases with interventricular septal defects ming and Stephenson s (1928) case showed complete heart block 70) Bundle branch block and left axis deviation was present in the

case of Calo (1937) The T waves may be increased in amplitude and a negative T wave of right ventricular hypertrophy was present in lead 2 in Segall s (1933) and other cases

RADIOLOGY There has been considerable divergence of opinion in regard to the radiological picture only clarified by increasing clinical experience Laubry and Pezzi (1921) stated that there was but little difference in the picture of pulmonary stenosis whether the septum was open or closed Laubry (1930) later thought that prominence of the





A B

FIG 71 The tetralogy of Fallot with right sided aortic arch

left middle arc was less frequent in the tetralogy than in isolated pulmonary stenosis Assmann (1928) noted a concavity at the site of the pulmonary arc and figured three cases with autopsy control Bedford (1929) stated that the characteristic radiological picture was that of an aorta projecting abnormally to the right with a notable concavity at the site normally occupied by the pulmonary arc the heart silhouette being of the coeur en sabot type Blackford (1930) likewise commented upon this concavity Papp (1931) considered the diagnosis to be essentially radiological and based upon the signs of Assmann and Bedford

It might be anticipated that a developmental abnormality involving hypoplasia of the pulmonary conus and artery would be accompanied by a concavity or at least an absence of the prominence caused normally by these structures This is in accord with the observed facts and the degree of concavity is perhaps in part determined by the severity of the pulmonary obstructive lesion The only exception to

such a statement is that occasionally the conus is sufficiently hyper trophied to form a bulge on the left cardiac border slightly lower than that of the pulmonary artery (Usomoto 1925) A similar case has been figured by Bedford (1929) In the case of White and Boyes (1932) a dilated actively pulsating pulmonary artery was observed during life and a diagnosis of patent ductus arteriosus with infective endocarditis was considered Autopsy revealed a typical example of the tetralogy with pulmonary arteritis which latter had lead to dilatation of the abnormal vessel If all these various points are taken into consideration it is understandable that some natural confusion may have arisen in regard to a completely characteristic picture of the tetralogy

The heart is most often normal in size or even smaller than normal and gross enlargement militates strongly against a diagnosis of the tetralogy It may be slightly enlarged to the left from right ventricular hypertrophy and then assume a silhouette with a blunt apex slightly raised above the diaphragm the coeur en sabot This picture occurs in but half of the cases A right sided aorta is present in about a quarter of the cases and its recognition may be of considerable surgical importance (fig 71)

The middle arc of the left cardiac border shows varying degrees of concavity this being most likely to be marked in pulmonary atresia or in high degrees of stenosis (figs 72 and 73) In some cases the left border may be almost straight and in an occasional case there may even be slight convexity of the pulmonary arc In the left anterior oblique position there is a large pulmonary window and hypertrophy of the right ventricle is obvious in comparison with the left In the right oblique view the concavity of the pulmonary arc becomes obvious

The condition of the lung roots is of the utmost importance Fluoroscopy shows diminished or absent pulsation In an occasional case it is possible to demonstrate more pulsation on the left side than on the right due to the presence of a patent ductus arteriosus Like wise in a few cases a well developed collateral circulation may give hilar appearances that are almost normal in regard to size but close scrutiny shows the shadows of a nodular appearance variable according to the development of the circulation A peppered appearance is rather characteristic of this contingency Corresponding to the diminution in the pulmonary blood flow there is an increased translucency of the lung fields a point of major importance Pulmonary congestion excludes the tetralogy There may be indentation of the barium filled oesophagus by the vessels of a collateral circulation

While it is possible in the large majority of cases to arrive at a correct diagnosis by these conventional methods it may be necessary to resort to angiocardiography in a proportion of cases This special method will give added information about the blood vessels that may

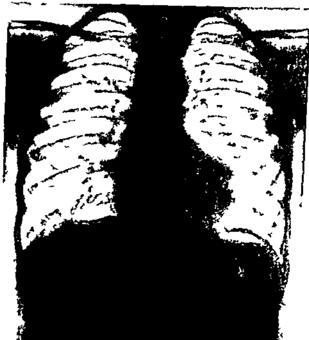


Fig 72 Pulmonary atresia Child aged 5 Pulmonary trunk a fibrous cord The patient died after a Pott's operation

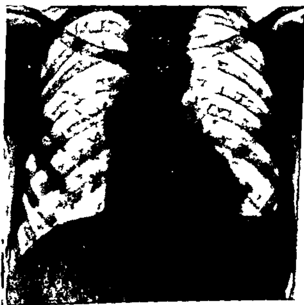


Fig 73 Tetralogy of Fallot Female aged 10 with

be used for anastomotic procedures The pulmonary artery can nearly always be seen except in atresia but there is delay in its filling dependent upon the severity of its stenosis A shunt from right to left fills the dextroposed aorta at two seconds The relative densities of the aorta and pulmonary artery shadows is a clue to the magnitude of this shunt and the severity of the stenosis The persistent translucency of the lung fields is the best indication of a defective pulmonary circulation The radiokymographic examination may also be valuable

COURSE AND PROGNOSIS Many cases die in infancy particularly those with pulmonary atresia where closure of the ductus before the development of an adequate collateral circulation may precipitate death There is a further peak of mortality at puberty Subsequent to this the numbers gradually diminish so that but few attain early adult life and fewer still reach a greater age

Bacterial endocarditis may claim a certain number of victims but its incidence is not high compared with other groups The sites especially involved are the conus of the pulmonary artery or the artery itself A personal case showed the pulmonary artery to be filled with vegetations distal to the pulmonary cusps and a similar case is that of White and Boyes (1932) The dextroposed aorta favours paradoxical embolism and death from post operative embolism of a pulmonary artery has been recorded Cerebral abscess has been the closing clinical picture in many cases the earliest being that of Farre (1814) Peacock thought that most congenital cyanotics died of some cerebral incident Abbott Lewis and Beattie (1923) collected eight cases and added one of their own Heart block is rare but occurred in the case of Nobecourt (1925) and Fleming and Stephenson (1928) Campbell and Susman (1934) refer to Moxon s case of pulmonary atresia where there was a pulse rate of 25 during life The rarity of heart block in the cyanotic case where the interventricular septal defect is unusually large has been a matter for comment Pulmonary tuberculosis is not uncommon despite frequent statements to the contrary Death may occur in an asphyxial attack or from intercurrent disease The average age at death where there is pulmonary artery hypoplasia is 12 years When pulmonary atresia is present life is generally shortened and the average age at death is 5 However the cases of East and Barnard (1938) lived to 20 33 and 30 years A few cases survive to later adult life the most remarkable being that of White and Sprague a noted musician who attained the age of 59 Brumlik s (1937) case survived to the age of 62

DIAGNOSIS The tetralogy has to be distinguished from other types of congenital heart disease accompanied by cyanosis For practical purposes it is possible to exclude those cases where cyanosis develops late or as a terminal event as in the interauricular septal defect the isolated patent ductus arteriosus or interventricular septal defect Certain other cases tend to be naturally excluded in the younger age

groups for the very complex primitive defects usually result in death before the age of three Unquestionably in the cyanotic congenital heart case surviving beyond the age of 3 the tetralogy is numerically preponderant but this does not preclude the necessity for accurate diagnosis if surgery is to be undertaken It has been stated that as high as 70 per cent of cyanotic cases surviving infancy are cases of the tetralogy

As a preliminary step the radiological examination is of the utmost importance because an enlarged pulmonary arc increased root shadows hilar pulsation and pulmonary congestion at once dismiss the tetralogy and imply a considerable pulmonary circulation Thus the Eisenmenger complex where a normal or dilated pulmonary artery replaces the hypoplasic vessel of the tetralogy is at once excluded

Transposition of the vessels may give some diagnostic difficulty Most cases die in early infancy but survival may be prolonged in those cases where there is an associated interventricular septal defect Cyanosis may be severe The physical signs are not distinctive The heart shadow has a globular contour and serial radiograms over a period of time show progressive enlargement of the heart with congestion in the lung fields The position of the aorta and pulmonary artery are the reverse of normal the aorta being anterior This presents a narrow vascular pedicle in the posterior anterior view and the position of the vessels relative to each other can be recognized in the oblique view

Tricuspid atresia presents a heart with a markedly concave pulmonary arc The most distinctive feature in diagnosis is that almost alone amongst cases of cyanotic congenital heart disease there is a left axis in the electrocardiogram

Cor biatriatum triloculare may include some cases of tricuspid atresia and it is rare for cases to survive into later childhood Apart from marked cyanosis the physical signs are not conspicuous There may be a systolic murmur but this is often in default and absence of a bruit might well be a pointer to some abnormality more complex and primitive than the tetralogy It has been stated that large biphasic ventricular complexes in all leads of the electrocardiogram are frequent in this general category of cases The heart is often enlarged and has not the contour of the Fallot

A persistent truncus arteriosus is usually accompanied by but slight cyanosis The cyanosis present depends upon the adequacy of the pulmonary branches arising from the aorta Physical signs are conspicuous there being generally a coarse systolic thrill and loud systolic bruit Hilar pulsation may be present or absent and the electrocardiogram commonly shows only a slight right axis or even a left axis

It may happen that in a few rare cases there may be pulmonary stenosis with a ventricular septal defect but without *dextroposition of* the aorta Such cases are cyanotic and may live to a considerable age

There is no certain method of identifying these cases and the X ray picture may well resemble that of the tetralogy Such a condition may be encountered at operation and it has been possible to enlarge the aperture in the stenosed area of the pulmonary artery

It is emphasized that the radiological picture of the tetralogy is generally quite characteristic and rarely leaves any doubt about a case In the few doubtful cases accessory methods of examination such as cardiac catheterization and angiocardiography may be necessary

SURGERY A new era in the treatment of congenital heart disease has opened with the introduction of a surgical treatment of the patent ductus arteriosus coarctation and pulmonary stenosis and atresia Few will doubt the wisdom of operation on the ductus in the young subject some may well question the validity of surgical intervention in coarctation none can doubt that any possible surgical ameliorative procedure is a necessary step in the management of the severe cyanotic case Such surgical treatment is all that is available to these unfortunate sufferers who may exist rather than live and it should be considered in every cyanotic case Accurate diagnosis is therefore essential and no method should be omitted in order that this may be achieved The surgery of cyanotic congenital heart disease affords a good example of purely physiological surgery designed to improve function in contrast to extirpative surgery which is concerned with the actual removal of diseased tissues and structures

It must not be thought that all forms of cyanotic congenital heart disease are suitable subjects for some type of ameliorative operation and so far the surgery of the cyanotic case can only be considered in relation to those abnormalities in which an inadequate volume of blood reaches the lungs for aeration Nor must it be considered that surgical operation is a cure of the abnormality for however markedly the cyanosis disappears there still remain the abnormalities of anatomical structure now with the additional factor of an artificially created ductus arteriosus prone to those complications to which a ductus is liable There is however to day the added protection of penicillin and other antibiotics

There are two possible surgical operations In the Blalock Taussig (1945) operation an anastomosis is made of the end of the subclavian carotid or innominate artery with the side of the pulmonary artery In the Potts Smith (1945) operation a direct side to side anastomosis is made between the pulmonary artery and aorta Both operations have their particular merits In the former procedure it is advantageous to use a subclavian vessel rather than the innominate although in very young subjects the latter may have to be used Mortality is less when the subclavian is employed The Potts Smith operation allows the operator to choose the size of the aorto pulmonary fistula that he creates Both operations succeed in their intent to furnish a more

adequate blood volume to the pulmonary circulation This is not the place to discuss actual surgical technique and for details the reader is referred to the original papers Suffice it to say that in the Blalock Taussig operation the incision is made in the third left space on the side opposite to that on which the aorta descends Despite the extreme cyanosis of the patient the operation involving the temporary shutting off of the circulation to one lung is remarkably well tolerated The Blalock Taussig operation is attended by an overall mortality of about 14 per cent Holmes Sellors (1948) and Brock (1948) have performed operations to divide an obstructing pulmonary valve in some cases of severe pulmonary stenosis Brock has further operated on infravalvular stenosis with success

Barrett and Daley (1949) have described a further method of increasing the blood supply to the lungs applicable to those cases where valvulotomy or anastomotic operations are impracticable or inadvisable Their operation aims at the production of vascular pleural adhesions and with this in view the pleural cavity is opened and the parietal pleura is removed from the upper mediastinum upper half of the chest and dome of the pleura Asbestos powder is then dusted on to the raw surfaces and the chest closed This operation is performed first on one side and at a later date on the other In another type of operation a pulmonary omentopexy has been done Such procedures have resulted in improved exercise tolerance diminution in cyanosis and other clinical improvements The arterial oxygen saturation has been increased in several of the operated cases

Effects of Operation It should not be supposed that the operation is a complete cure of the condition for the anomalous structural changes still remain with the addition of an artificial ductus arteriosus The immediate effects of operation are a great clinical improvement in the patient There is a marked decrease in polycythaemia with an increased oxygen saturation of the arterial blood This latter never reaches the normal level of 96 per cent because of the overriding aorta but figures from 70 to 85 per cent are attained Thus children who could only walk a few yards and then with great distress may walk for miles after the operation

Blalock (1947) has operated upon 474 patients with eighty six deaths from all causes including errors in diagnosis The table gives his results according to the vessel employed to make the anastomosis In not one of the cases where the subclavian was used has there been any difficulty with the circulation in the arm Neither has the use of the innominate artery with its implied ligature of a common carotid been as permanently prejudicial to the cerebral circulation as might have been anticipated Blalock states that weakness or paralysis of the contra lateral side of the body has disappeared or largely cleared up in all cases that survived operation and cerebral lesions due to ischaemia

have been very rare This is in contrast with the permanent hemiplegias that occur spontaneously in the cyanotic case The creation of an artificial ductus arteriosus has not so far resulted in the perils of infective endocarditis cardiac hypertrophy or premature cardiac failure Penicillin and other antibiotics have so materially improved the prognosis of bacterial endocarditis that the risk may be almost negligible Lastly the thought must occur as to whether there is so far any substantial evidence that the anastomotic opening will grow and keep pace with the growth of the individual There must be considerable scarring around the anastomosis and this might well be prejudicial to the continuance of an adequate shunt In this connection it may be recalled that regression of the isolated interventricular septal defect may perhaps be due to contraction of its fibrous margins so sealing it off The recent work of Campbell and Susman (1947) relative to coarctation and the development of a collateral circulation suggests that the actual size of coarctation does not increase in diameter at the same rate as the rest of the aorta if at all so that relatively the degree of stenosis *vis à vis* the aorta increases until the age when full physical development has been completed If this concept is true then the artificial ductus created by operation may not be adequate at a later age The correct answers to all these questions will only be available when time has elapsed and a long series of operated cases can be assessed It can be said of the Potts Smith (1946) operation that the surgeon can choose the size of the anastomotic opening that he thinks fit and there is no possible danger to the cerebral circulation The disadvantage is that too large an anastomotic opening might result in cardiac hypertrophy and dilatation and early death Potts and Gibson (1948) state that there has been some enlargement of the heart in every case owing to the extra work created in the artificial ductus This becomes stable in a month or two and although they are fearful in a few cases (three) no heart failure has appeared

They have operated on forty five patients with four deaths a mortality of 8 8 per cent

Selection of Cases for Surgery The diagnosis of the tetralogy or of some congenital abnormality involving a deficient blood supply to the lungs must be reasonably certain The best time for surgery is between the ages of five and ten years but the operation may be done below the age of five in cases where necessity arises owing to marked deterioration of the patient The difficulties of operation appear to increase with age It is necessary that there should be both systemic and pulmonary vessels suitable for anastomosis Indications for urgent operation are a rising blood count spontaneous attacks of dyspnoea and deterioration of the patient as a result of anoxaemia Contra indications to operation are gross disturbances of conduction evidence of bacterial endocarditis renal disease or recent cerebral accidents Opinions

might conflict as to the desirability of operating on a case where a ductus arteriosus remains patent owing to the alterations in pressure gradients occasioned by operation

TABLE SHOWING RESULTS OF OPERATION ON CYANOTIC CASES
BLALOCK INTERNATIONAL CONFERENCE PHYSICIANS LONDON 1947

ANASTOMOSIS	*Cases*	*Deaths*	*Per cent*
Subclavian and Pulmonary Artery			
End to side	331	37	11
End to end	23	4	17
Carotid and Pulmonary			
End to side	30	9	30
End to end	*1*	*1*	*100*
Innominate and Pulmonary			
End to side	47	13	28
End to end	1	1	100
Aorta and Pulmonary			
Side to side	2	1	50
Exploratory thoracotomy	37	18	49

The Eisenmenger Complex

The essential features of this anomaly are a high defect of the inter ventricular septum dextroposition of the aorta and a normal or dilated pulmonary artery The condition is held to be a relatively rare one and was so named by Abbott after the case described by Eisen menger (1897) The earliest case however is that of Dalrymple (1847) *It has often been considered as a type of the tetralogy of Fallot* but this is inaccurate Its distinctive distinguishing character is a normal or dilated pulmonary artery in contrast to the invariable pulmonary stenosis or atresia of the tetralogy

PATHOGENESIS The general appearance of the heart suggests that the conus and pulmonary artery have developed normally and that the endocardial cushions of the atrial canal have in the main fulfilled their correct functions There is a large septal defect at the base of the inter ventricular septum and Rosedale (1935) considered that failure of the lower part of the bulbar ridges to fuse and consequent failure of the aortic vestibule to be shifted to the left resulted in dextroposition of the aorta A shunt from left to right through the defect would result in dilatation of the conus and pulmonary artery Others have held different views *Streptococcal endarteritis was thought to be the cause* of pulmonary artery dilatation in the case of Blechman and Paulin (1922) Stewart and Crawford (1933) adduced histological evidence of a syphilitic process as a possible cause of the cardiac anomalies with secondary pulmonary arteritis to explain pulmonary artery dilatation Taussig and Semans (1940) case also presented old healed infective lesions The balance of evidence suggests that the presence of infective processes is the result of the congenital lesion rather than the cause

ANATOMY There is a high ventricular septal defect anterior to the

membranous septum and it is of similar character to and has the same smooth fleshy margin as in the tetralogy (fig. 74) The aorta is displaced to the right to a variable extent and overrides the septal defect The course of the aorta is normal as a rule but a few cases show a right aortic arch It is not uncommon to find deformity of the aortic cusps with accompanying aortic incompetence The cusps may be unequal in size the cusp immediately above the septal defect being the largest The pulmonary artery may be normal in size or larger than



Fig 74 The Eisenmenger complex Child aged 2 died of pneumonia Pulmonary artery enlarged and the aorta lying astride a high ventricular septal defect

normal and is found in its usual position The right ventricle shows signs of hypertrophy and dilatation and similar changes of lesser degree may be present in the left ventricle The conus of the right ventricle is often large and thick walled Calcification may be present in the pulmonary cusps as in the case of Stewart and Crawford and in the left pulmonary artery in an unpublished case of Hubble

Bacterial endocarditis of the pulmonary valve and pulmonary arteritis occurred in the cases of Abbott (1927) Millman and Kornblum (1936) and Bedford and Parkinson (1936) In the latter case there was a right aortic arch In the case of Talley and Fowler (1936) there was a general hypoplasia of the aorta involving its entire length Cases with slight dextroposition may be indistinguishable from the isolated ventricular septal defect

CLINICAL PICTURE The factors in the production of cyanosis are a shunt of mixed blood into the overriding aorta and failure of adequate oxygenation in the lungs This latter may possibly be due to changes in the lungs induced by an excessive pulmonary blood flow or as thought by some due to congenital abnormality of the pulmonary endothelium In the bulk of cases cyanosis is minimal or absent for many years It tends to appear towards puberty or later, and is moderate in amount with negligible clubbing Cyanosis may also be episodal and related to exertion emotion or intercurrent infection Perhaps too the age at which cyanosis appears may be determined by the degree of dextroposition of the aorta for if it lies nearly wholly in the right ventricle the chances of cyanosis are greater than when dextroposition is only slight in degree Dyspnoea is always more obvious than cyanosis in the early stages Haemoptysis may occur Baumgartner and Abbott s (1929) case showed hoarseness culminating in aphonia due to pressure on the recurrent laryngeal nerve by the hypertrophied conus of the right ventricle The heart is only slightly enlarged There is often pul sation in the pulmonary area with a diastolic shock indicative of pul monary artery dilatation A harsh systolic murmur is characteristically loudest in the pulmonary area It may be widely audible over the prae cordium and be transmitted downwards to right and left It is not heard in the neck vessels but may be heard in the interscapular region A diastolic bruit of pulmonary incompetence may be present Aortic incompetence may be associated Both systolic and diastolic thrills have been reported

RADIOLOGY The characteristic feature of the radiological picture is a greatly enlarged pulmonary arc due to hypertrophy of the conus and dilatation of the pulmonary artery (figs 75 and 76) The heart may have a globular outline in young children In older cases the apex is blunt and raised above the diaphragm The hilar shadows are increased owing to the dilated pulmonary branches and vigorous pulsation is present on the screen The shadow of the aorta is more to the right than is normal The picture may resemble that of the interauricular septal defect but there is not the gross enlargement of the latter abnormality As in other cases of cyanotic or other congenital heart disease the radiological examination plays an integral part in diagnosis Likewise as in other forms of congenital heart disease it is possible to have a radiological picture that is not truly characteristic and which is of little help in



(a) Posteroanterior



(b) Left anterior oblique

Fig 75 Eisenmenger complex

diagnosis The case of Millman and Kornblum (1936) a female aged 32 showed no dilatation of the pulmonary artery

ELECTROCARDIOGRAM Right axis deviation of varying degree is nearly always present Conduction disturbances are rare Bundle branch block occurred in the cases of Moffatt and Eakin and Baumgartner and Abbott Right axis with runs of dissociation was a feature of the clinical case of Glazebrook (1943)

COURSE AND PROGNOSIS Many of the reported cases have attained



FIG 76 The Eisenmenger complex in a child aged 12 The heart has a globular outline and the aorta is dextraposed The pulmonary artery is dilated

middle life without disability Despite a life of dissipation Stewart and Crawford s case lived to the age of 60 Death may be due to cardiac failure cerebral abscess or intercurrent infection The average age at death in Abbott s series was 16 years The outlook is not good in those cases that develop cyanosis early

DIAGNOSIS The salient features of the Eisenmenger anomaly are that cyanosis tends to appear late and the radiological picture of an enlarged pulmonary arc with pulsating pulmonary arteries is characteristic Undoubtedly the diagnosis may be very difficult at times and only arrived at by a process of elimination in which all the accessory methods of examination may be necessary The presence of a venousarterial shunt may be demonstrated by a reduced circulation time which will at the same time tend to exclude the diagnosis of isolated pulmonary stenosis or isolated ventricular septal defect both of which may present a comparable clinical picture In a frankly cyanotic case the

diagnosis of the tetralogy is vitiated by the prominent pulmonary arc and hilar pulsation In childhood an auricular septal defect may be suspected but the Eisenmenger anomaly is not accompanied by the small aorta of that condition Angiocardiography and cardiac catheterization have a definite place in the diagnosis of this abnormality Angiocardiography shows simultaneous filling of both aorta and pulmonary trunk and dense pulmonary vascular markings Cardiac catheterization shows elevation of pressures in the right ventricle and pulmonary artery The oxygen content of right ventricular blood is markedly higher than that from the right auricle

(fig 77) Atresia is present at the origin of the aorta and hypoplasia involves the vessel as far as the entrance of the ductus arteriosus The coronary arteries arise at their usual situation just above the atresia This latter may be either a thin membrane or there may be evidence of fused cusps Occasionally it is a thick mass of tissue In inflammatory cases there may be a minute passage from the left ventricle to the aorta The pulmonary artery is enlarged and continuous with a widely patent ductus arteriosus The left ventricle may exist as a small cavity in the



FIG 78 The heart in aortic atresia Note the thickened pearly endocardium of the left ventricle

wall of the right ventricle and careful examination ought to be made before concluding that there is a common ventricle In other cases there is a ventricular septal defect communicating with a small left ventricle In some inflammatory cases the left ventricle has been hypertrophied and dilated The pearly white endocardium of endocardial thickening may be present in both the inflammatory and the developmental types It appears from the published cases that the ventricular septum can be entirely absent or it may be defective or quite normal Coarctation was present in a case of Evans (1933) and in the case of Dreyfuss (1929)

CLINICAL PICTURE Extreme cyanosis with dyspnoea is the rule This may not be immediately evident at birth but develops within a few hours In cases with extreme aortic stenosis cyanosis is present but

not so marked as in the case of true atresia Most often there are no abnormal auscultatory signs but there may be occasionally a systolic murmur

COURSE AND PROGNOSIS Life is usually only a matter of a few hours Summon s (1906) case lived for fifteen weeks

Mitral Stenosis and Atresia

The mitral valve may be narrowed giving rise to a mitral stenosis which if congenital is always accompanied by secondary hypoplasic changes in the left side of the heart and aorta In many cases changes in the valves are in the nature of an aplasia and the chordae and papillary muscles are present In certain instances there has been evidence of an inflammatory process as the probable cause as distinct from a purely developmental etiology When the mitral valve is com pletely obliterated mitral atresia is said to be present and is almost invariably a developmental anomaly In mitral atresia the valves and their appendages may be entirely absent and there may be no trace whatsoever of the left auriculoventricular orifice A severe mitral *stenosis may amount to a functional atresia and consequently there* is some overlapping of the two conditions

The mitral valve is differentiated from the endocardial cushions of the atrial canal As development proceeds the cushions grow down wards and meet the muscular trabeculae of the ventricular wall Further changes take place whereby the musculi papillares chordae and cusps are evolved During the phase of evolution growth may become arrested and the further changes taking place in the heart are largely in response to the altered hydrodynamic condition A narrowed mitral valve entails a diminished amount of blood passing to the left ventricle and aorta and if stenosis is severe hypoplasia of these structures follows Correspondingly an increased blood flow in the right side of the heart leads to hypertrophy and dilatation of the right chambers The ductus may remain patent and continuous with the descending aorta Little is known of the actual factors responsible for the arrest of development and aplasia of the mitral valve The usual integrity of the interventricular septum suggests that its occurrence may be after the eighth week in many cases About half of the recorded cases appear to be actual aplasia of the mitral valve the others showing definite evidence of a foetal endocarditis Field (1938) has collected seven cases of congenital mitral stenosis largely in male infants from the post mortem records of the Great Ormond Street Children s Hospital Three of these cases showed aplasia of the left auricle and ventricle In one case a patent ductus arteriosus was present In three cases there was a defect of the interventricular septum and in two cases tricuspid stenosis was associated Both cases with tricuspid stenosis showed recent or old endocarditis of the mitral valve In at least three

of the other reported cases (Fischer 1911 Kockel 1934 and Farber and Hubbard 1933) the lesion was attributed to a foetal endocarditis owing to disease of the mother in the later stages of pregnancy Each of these cases had additional aortic stenosis and the histological evidence was *confirmatory of an infective etiology* It thus seems possible to recognize a purely developmental hypoplasia and an infective type both resulting in various degrees of abnormality of the mitral valve

'Pure Mitral Stenosis

In view of the controversy which has raged for many years concerning pure mitral stenosis this lesion is briefly considered here The term *pure designates a type of mitral stenosis occurring in young subjects* in whom there are no signs of incompetence and a complete absence of a rheumatic history Included in this category may be some cases of Lutembacher s disease (Chapter X) where mitral stenosis is associated with an auricular septal defect In these circumstances mitral stenosis has been held to be a part of a more complex congenital abnormality involving disproportion between the aorta and pulmonary artery Weill (1895) and Huchard (1905) contended that pure mitral stenosis *was of developmental origin and referred to the condition as the* maladie de Duroziez in contradistinction to mitral stenosis of rheumatic origin the maladie de Vieussens so named after the classical description of rheumatic mitral stenosis by Vieussens in 1715 Vaquez (1924) commented that none of the protagonists of a developmental mitral stenosis had ever satisfactorily demonstrated the condition in an autopsy of a newborn infant The conception of pure mitral stenosis still lingers in France although increasing knowledge of the rheumatic *infection* has gradually displaced the idea of a congenital etiology of mitral stenosis in cases without rheumatic antecedents Analogy may be found in the concept pure aortic stenosis again largely recognized by French clinicians *If it is admitted that these cases of aortic stenosis* are the result of an infective process installing itself in earliest infancy it seems likely that some cases of mitral stenosis may arise in the same way The relationship of mitral stenosis to the auricular septal defect is discussed elsewhere

Indubitably there are a few cases of congenital mitral stenosis as autopsy records show (Donnally 1924 Field 1938) but these are rare and are more often combined with other congenital cardiac abnormalities *The presence of extracardiac malformation in a case of mitral* stenosis might evoke suspicion particularly in a young child of a congenital cardiac lesion Such cases have been reported and the association laboured but it is unwise *to draw any firm conclusion The* Lutembacher syndrome is well known and its examples are conspicuous *by the frequent absence of* any rheumatic history (McGinn and White

1933 Bedford Papp and Parkinson 1941) The work of Roesler (1934) and others and more recently the review of Burrett and White (1945) has emphasized that the mitral valves in cases of auricular septal defect are peculiarly susceptible to infection and for this reason alone it is unwise to always assume a congenital origin of the mitral stenosis present

Donnally s (1924) case may be taken as the type of congenital mitral stenosis There was a funnel shaped baggy valve undifferentiated into cusps and its orifice measured 3 mm Papillary muscles and chordae *were present The cavity of the left ventricle was rudimentary* and marked hypoplasia of the aorta was present The foramen ovale was open and the ductus arteriosus widely patent and continuous with the descending aorta This case exemplifies the marked changes in the left heart due to deprivation of the blood flow A congenital mitral stenosis is figured by Huchard (1905) and the valve is described by him as resembling a tunnel Its orifice measured 7 mm and viewed from the hypertrophied left auricle the endocardium of the orifice was folded and resembled the anus It appears that the recognition of a congenital mitral stenosis is largely in the post mortem room and is dependent not only upon the changes in the valve itself but also upon the secondary changes in the left ventricle and aorta Lastly mention might be made of those cases of mitral stenosis associated with congenital syphilis Raynaud *et al* (1939) mention twenty four such cases

CLINICAL PICTURE Donnally reviewed twelve cases from the literature Cyanosis and dyspnoea coming on shortly after birth were present in all the cases Cyanosis was present in four of Field s cases Clubbing may be present in those cases that survive sufficiently long A definite presystolic murmur was present in two of Field s cases and a systolic murmur characterized as loud or blowing was present in the others Eight of Donnally s cases died within two months and the eldest survived to the age of 3 5 years In Field s series the average age at death was 6 months with extremes of 4 days and 1 75 years Death is usually sudden

Mitral Atresia

Mitral atresia is a quite rare congenital abnormality and only five examples are included in Abbott s 1 000 cases Manhoff and Howe (1945) mention twenty eight reported cases and add one of their own

Atresia of the mitral orifice may be complete and lead to its actual obliteration or a stenosis of the orifice may be so severe as to amount to functional atresia The term mitral aplasia sometimes applied to these cases is misleading because it is contrary to the actual process which results in the anomaly Aplasia should only apply to those cases where the mitral valves are small and underdeveloped and the valve incompetent

In complete atresia arrest of development takes place at an earlier stage than in stenotic lesions In support of this there has been complete absence of the interventricular septum in a few cases It has been supposed that during the division of the atrioventricular canal by fusion of the anterior and posterior endocardial cushions the cushions may be deviated to the left and become adherent to the left wall of the common orifice thus causing atresia The process is one of hypertrophy of the cushions and this perhaps in some instances may be initiated by infection at the earliest stage Abbott (1927) suggests that lack of or exaggeration of the normal shift to the right of the common orifice might result in atresia of either the mitral or tricuspid valve She concluded that the probable explanation of the anomaly is a malposition and irregular union of those parts of the cardiac septa separating the mitral from the tricuspid ostium Undoubtedly in those cases where there is associated aortic atresia it is possible that a misplaced ventricular septum blocking the aortic ostium might initiate a process of left sided hypoplasia and be the primary event Monckeberg (1924) considered that unequal division of the truncus arteriosus might also account for some cases of mitral and aortic atresia Against the hypothesis of a malposed interventricular septum is that in some cases of mitral atresia the interventricular septum is entirely absent and in any event it is after the mitral and tricuspid orifices have been formed that the interventricular septum comes into contact with the fused anterior and posterior endocardial cushions of the atrial canal Harris and Farber (1939) studied mitral atresia in relation to transposition of the vessels and of twenty five collected cases fourteen presented some form or other of transposition On the basis of Spitzer s theory dextro position of the aorta might well be due to a diminished blood flow to the left ventricle and an increased flow to the right ventricle keeping the right sided aorta open and allowing the left sided aorta to close and disappear The time at which mitral atresia took place would determine the position and size of the aorta If before obliteration of the right sided aorta a normal sized or atretic dextroposed aorta will result if after obliteration of the right sided aorta then a normal sized or atretic aorta in its normal site will result This is in accord with the observed cases but does not explain those cases where there is complete transposition of the vessels a circumstance accepted by Manhoff and Howe (1945) as coincidental

Mitral atresia is usually associated with other defects of the heart and great vessels In one group of cases there is aplasia of the left ventricle and hypoplasia of the aorta Hypoplasia of the aorta may be extreme to the point of atresia In other cases there is dextroposition or even complete transposition of the great vessels

The anatomical features of the lesion reflect the altered circulation in the heart The stream of blood entering the left auricle is deflected

through an interauricular septal defect into the right auricle Often the left auricle is small and hypoplasic and is little more than the expanded continuation of the pulmonary veins The left ventricle may exist simply as a functionless cleft in the wall of a large common ventricle Or if a great vessel arises from this hypoplasic chamber there is an interventricular septal defect The larger common ventricle is hyper trophied and dilated The mitral valve and its appendages are usually entirely absent If present the cusps are small fused and undifferentiated and the chordae may be lacking The coronary supply in the case of the non transposed aorta may be poor owing to aortic atresia and the necessity for blood to reach the aorta through a patent ductus arterio sus The general picture is that of a functional cor triloculare

The association of mitral and aortic atresia has been reported by Monckeberg (1924) Dolgopol (1934) Sprenkel (1936) and Walker and Klinck (1942) Such cases are similar to but the reverse of pulmonary and tricuspid atresia The coronary arteries receive blood from the cul de sac of the ascending aorta The case of McIntosh (1926) is of especial interest in that the foramen ovale was not patent The only route by which aerated blood reached the heart was through a right pulmonary vein which entered the superior vena cava and an abnormal vessel which ran from the left auricle to the superior vena cava The ductus arteriosus was patent

There is no means of recognizing these cases during a life which is inevitably brief Most cases die in the first few days of life but some cases with transposition of the vessels may live for a few months

Tricuspid Atresia

Tricuspid atresia is a relatively infrequent congenital abnormality Manhoff and Howe (1945) found thirty three cases in the literature and added one of their own To these may be added the cases of Sabrazes and Hervé Cras (1941) Cathala (1942) Dustin and Denolin (1946) and Dustin Henry and Dustin (1947) These latter estimate that about fifty cases are known When the rather characteristic physical signs and laboratory findings are more fully appreciated it seems likely that more of these cases will be recognized

PATHOGENESIS Various theories have been advanced to explain tricuspid atresia which bears a close resemblance to mitral atresia Unequal division of the common auriculoventricular orifice by fusion and adhesion of the anterior and posterior cushions to the right wall has been suggested Abbott s opinion is embodied in her statement that the abnormality arises in malposition and irregular union of those parts of the cardiac septa dividing the mitral from the tricuspid ostium Chronologically such an event occurs at about the fourth week of intra uterine life At this stage the anterior and posterior cushions of the atrial canal have fused except at their right and left extremities These

extremities are destined to become the right and left auriculoventricular ostia and by thickening of the endocardium right and left endocardial cushions are formed from which develop the tricuspid and mitral valves The developing interauricular septum joins with the middle part of the fused anterior and posterior cushions and the developing interventricular septum joins the right posterior endocardial tubercule causing some narrowing of the right ostium This is only temporary for later the interventricular septum is rotated so that its anterior part

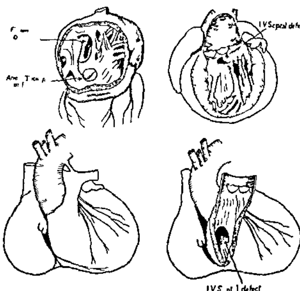


FIG 79 Diagrams of the heart in tricuspid atresia with an interventricular septal defect Child aged 8 months

comes to the right and its posterior part to the left This rotation brings it into alignment with the atrial septum It seems likely that in tricuspid atresia normal rotation of the interventricular septum fails and the right ostium is compressed and finally obliterated The associated ventricular septal defect is a true defect and represents failure of union between the bulbar septum and the ventricular septum owing to lack of rotation Similarly the frequent coincidence of pulmonary atresia and pulmonary conus stenosis suggests failure of torsion and unequal division of the bulbus and truncus Tricuspid atresia has sometimes been ascribed to the effects of a foetal endocarditis and occasionally a case may be seen in which there are rudimentary and fused valve leaflets Much more often the site of a tricuspid valve is a smooth fibrous membrane or trabecular muscular tissue in which no suggestion of valvular structure can be found The occurrence of dextroposition

or of complete transposition in many cases lends strength to a developmental origin

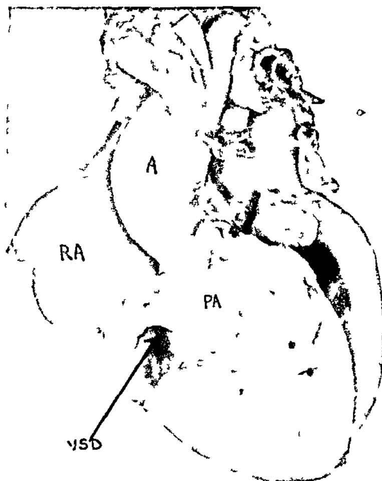
Assuming the atresia to arise at about the fourth week such abnormalities as the accompanying patent foramen ovale or persistent ostium secundum patent ductus arteriosus hypoplasia of the pulmonary tract are really secondary abnormalities determined by the need of a circulation to the lungs on the one hand and by altered dynamic changes in the blood flow on the other The extent of these secondary defects determines whether the heart will function as a biloculate or triloculate heart

ANATOMICAL FEATURES The distinction between a developmental and an infective etiology is usually apparent on microscopical examination if not on the naked eye appearances Atresia the result of infection is shown by scarring of the cusps and of the adjacent endocardium and by the presence of chordae and papillary muscles in the right ventricle In the developmental cases the appearances are quite different and cases may be divided into two groups those where there is an isolated atresia and those where there is some anomaly of the great vessels such as dextroposition of the aorta or complete transposition In Brown s (1936) case an elliptical depression marked the site of the tricuspid valve (figs 79 and 80) The right auricle was hypertrophied and the right ventricle existed as a small cavity the size of a hazel nut There was no trace of the valve appendages in the right ventricle The left ventricle was hypertrophied and there was a small interventricular septal defect beneath the non-coronary cusp The pulmonary artery itself was thin walled though of normal calibre and arose from the diminutive cavity The pulmonary conus was diffusely narrowed This case is an example of that group in which there is a small blood flow through a narrowed pulmonary tract In an unpublished case of tricuspid atresia and pulmonary atresia there was no communication between a pulmonary artery that existed as a fibrous cord and the right ventricle blood reaching the lungs through a patent ductus arteriosus A similar case is that of Holder and Pick (1939) In the case of Manhoff and Howe (1945) there was frank dextroposition of the aorta and complete pulmonary atresia co existed Dustin and Denolin (1946) reported a case with complete transposition of the great vessels the aorta in this case springing from a slit like right ventricle

A complete transposition of the vessels is a favourable circumstance as shown by the case of Hedinger (1915) a woman who led an active athletic life and survived until the age of 56 Presumably prolongation of life was due to the position of the pulmonary artery in the left ventricle which facilitated an adequate pulmonary blood supply In this case cyanosis was not a prominent feature until the terminal stage Cathala and Tisserand (1926) have reported the only case in which there was no interventricular septal defect

In tricuspid atresia it is evident that a reduced pulmonary blood flow may be the result of either a narrow ventricular septal defect narrowing of the pulmonary conus or actual atresia of the pulmonary artery Even when the vessels are transposed there may be pulmonary artery narrowing or atresia In the case with pulmonary atresia the circulation to the lungs is maintained through a patent ductus arteriosus

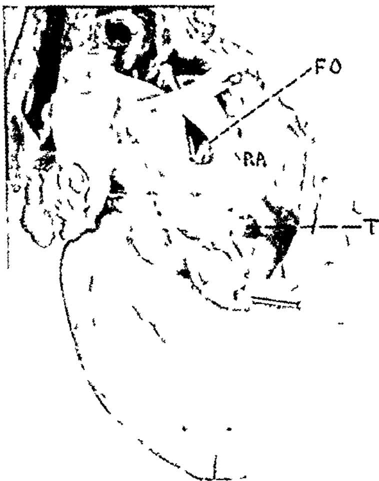
These anatomical findings are of importance in regard to possible surgical treatment In those cases where there is simple obstruction to the pulmonary flow either by conus stenosis or a small ventricular



(a) The aplastic right ventricle has been opened and an arrow points to a ventricular septal defect

FIG 80 Tricuspid atresia Female child aged 7 months

septal defect there must exist a low pressure in the pulmonary artery and therefore such a case is likely to benefit by an anastomotic operation In the other type of case where a pulmonary circulation is assured by a patent ductus arteriosus there will be a higher pressure system in the pulmonary artery and it is dubious whether there would be a sufficient pressure gradient to assure a flow through an artificially

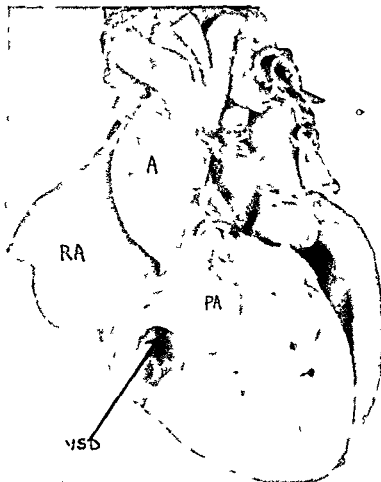


(b) Interior of the right auricle Depression at site of the tricuspid valve Foramen ovale open

markedly cyanotic with a normal axis in the electrocardiogram

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(a) The aplastic right ventricle has been opened and an arrow points to a ventricular septal defect

Fig 80 Tricuspid atresia Female child aged 7 month

inevitably have pulmonary stenosis and a defect allowing a shunt has perhaps contributed to this As some of the cases survive for a considerable period and are possible candidates for surgical relief the clinical features are worth careful consideration The presenting feature is cyanosis of variable degree with corresponding clubbing of the fingers in the older cases Cyanosis generally exists from birth and is enhanced by movement sucking or emotion but cases are on record such as the recent case of Dustin Henry and Dustin (1947) where cyanosis was

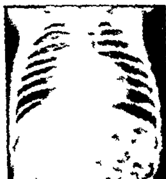


FIG 82 Tricuspid atresia Child aged 7 months

not evident until the child was several weeks old Cyanosis is likely to be late in a case with transposition of the vessels There may be frequent attacks of paroxysmal dyspnoea which come on quite suddenly last an hour or two and then cease spontaneously These attacks are not relieved by the inhalation of oxygen

Polycythaemia is always present The heart is enlarged to both the right and left and the apex beat is forcible A systolic murmur may be present in the bulk of cases and is best heard in the second and third left space close to the sternum It may only be present from time to time and not necessarily constant to the individual case Taussig (1936) states that it is due to flow in a patent ductus arteriosus It might also arise at the interventricular septal defect During attacks of cyanosis accompanied by severe dyspnoea the murmur may disappear In a few cases no murmurs have been heard at any time

RADIOLOGY The X ray picture discloses a rather globular enlarged heart The convexity of the right border corresponding to the right auricle is exaggerated The left border is well rounded and has the appearance of left ventricular hypertrophy (fig 83) Owing to absence of the pulmonary conus there is a concave pulmonary arc In the left anterior oblique position (fig 84) the right ventricle is seen to be very

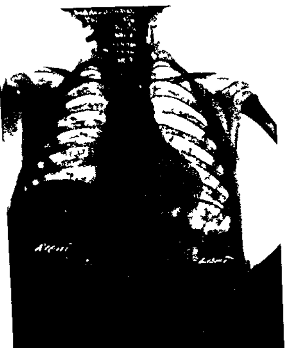


Fig 83 Tricuspid atresia Female aged 7½ years



Fig 84 Tricuspid atresia Female aged 2½ years Left anterior oblique view shows enlargement of the left ventricle E C G left axis

small and the left is large (fig 82) Angiocardiography shows a small right auricle The contrast medium enters the left ventricle in about two seconds without passing through the right ventricle showing that its only route is through an auricular septal defect The aorta and its branches are visible in the three second film A little later the right ventricle and pulmonary artery shadows appear the latter never dense



FIG 85 Electrocardiogram in tricuspid atresia

ELECTROCARDIOGRAM Almost alone amongst the permanently cyanotic group the electrocardiogram shows a left axis (fig 85) The tetralogy of Fallot with dextrocardia also shows a left axis but the P wave is inverted in lead 1 A few cases of persistent truncus or of common ventricle also show a left axis

DIAGNOSIS Cyanosis with paroxysmal dyspnoea clinical and X ray signs of left ventricular hypertrophy and a left axis in the electro cardiogram should be adequate grounds for diagnosis Cardiac catheterization would prove the presence of an auricular septal defect and it would not be possible to enter the right ventricle The angiocardiogram is discussed above

PROGNOSIS The average age at death in Abbott s series was 5 years with an extreme of 56 It is probable that this figure is low as recent work suggests that a number of cases survive to adolescence The two cases of Geraci Dry and Burchell (1948) were well and alive at 35 The prognosis depends upon the effectiveness of oxygenation in the lungs and the volume of blood reaching the lungs for aeration

Congenital Pulmonary Insufficiency

Congenital pulmonary insufficiency may occur in those cases where there is a communication between the two sides of the heart and is most frequently encountered in auricular septal defects and in the patent ductus arteriosus In both of these conditions the pulmonary artery is dilated and there is commonly in the case of the former an

associated hypoplasia of the aorta Pulmonary insufficiency as an isolated anomaly is exceedingly rare and it may arise in several ways Unequal division of the common arterial trunk has been described as a cause Defective formation of the pulmonary artery or an absent or defective cusp may also give rise to insufficiency In another group ante natal endocarditis involving the pulmonary valve may give rise to stensosis and incompetence Despite the not infrequent finding of a pulmonary valve of funnel shaped appearance owing to fusion of the cusps and the apparent impossibility of such a valve to completely close a diastolic murmur is rarely heard Another underlying cause of pulmonary insufficiency is a supernumerary pulmonary cusp Kissin (1936) states that in the literature there are 154 cases of a super numerary pulmonary cusp and in three of these there was definite pulmonary regurgitation and in seven additional cases autopsy showed that there was pulmonary incompetence The anatomical features of the supernumerary cusp vary from case to case and in many cases it is deformed and fenestrated

CLINICAL FEATURES The clinical signs are a diastolic murmur in the pulmonary area and along the left sternal border unaccompanied by the changes in blood pressure or peripheral signs of aortic incompe tence The heart may be slighly hypertrophied but gross enlargement is unusual The X ray shows a mitral configuration with a prominent pulmonary arc and this is well shown in Kissin s case

COURSE AND PROGNOSIS The average age at death is 40 with ex tremes of 5 months and 80 years

DIAGNOSIS The chief difficulty in diagnosis lies in the resemblance of the heart signs to the Graham Steell murmur of mitral stenosis and the similarity of the radiological picture to that of mitral stenosis Differentiation is possible in that the signs of mitral stenosis are absent there is no history of rheumatism and acquired disease of the pul monary valve is exceedingly rare The murmur of aortic incompetence can be differentiated by the absence of a collapsing pulse

Congenital Tricuspid Insufficiency

Congenital tricuspid insufficiency is a more serious condition than pulmonary insufficiency and may occur alone or in association with tricuspid stenosis

Tricuspid stenosis and atresia have been discussed above and their origin as the result of inflammatory or developmental processes con sidered There are other anomalies of the tricuspid valve Incomplete differentiation of the cusps may occur There may be an increase in the number of cusps or the cusps may be involved in a congenital hypo plasia (Ariel 1930) The cusps may be reduced in number two only being present in the cases of Hotz (1923) Palladino and Kinney (1948) have described two cases one aged 34 in which the septal cusp was

adherent to the ventricular wall by bands of endocardial tissue and abnormally short chordae tendineae were present A displacement of the valve downwards into the right ventricle was described by Ebstein (1866) and this generally occurs in an otherwise normal heart It is a rare abnormality and there are twelve reported cases The morbid appearances of such a heart are striking because the line of attachment of the valve is roughly vertical instead of its usual transverse lie The result is that the right side of the heart is so divided that a large part of the right ventricle functions as a part of the right auricle The anterior cusp always retains its attachment to the annulus fibrosus at any rate in part and is usually the largest cusp present The posterior cusp has usually no connection with the annulus but is attached to the walls of the ventricle The septal cusp tends to be small and under developed Consequent upon the abnormalities of the cusps there are variations in the position and size of the papillary muscles of which some may be absent and chordae only present The true auriculo ventricular orifice is dilated and the malformed valve incompetent The right auricle and part of the right ventricle are grossly dilated The pulmonary conus and artery may be likewise dilated A patent foramen ovale has been associated in all but two of the reported cases but other anomalies other than those of the venous valves are rare A recent case is that of Brekke (1945)

CLINICAL PICTURE The clinical features are of interest and vary to a certain extent with the underlying cause of insufficiency In the Ebstein anomaly cyanosis may depend upon the amount of blood passing through the foramen ovale and upon the volume of blood entering the pulmonary artery Cyanosis may thus be only slight although it has been marked in certain of the reported cases Clubbing depends upon the degree of cyanosis present The heart is enlarged and there is often a loud systolic murmur in the third and fourth left spaces close to the sternum A presystolic or diastolic murmur may be heard at the same site A murmur was not present in Brekke s (1945) case A thrill may be present Extrasystoles and paroxysmal tachycardia are common At a later stage when failure develops the classical signs of tricuspid incompetence appear There are distended neck veins an enlarged and pulsating liver and passive congestion of the viscera Hotz (1923) described weakness of the left pulse due to pressure on the aortic arch by an enlarged pulmonary artery and paralysis of the left recurrent laryngeal nerve for a similar reason

In tricuspid insufficiency due to causes other than that of the Ebstein type the duration of life is usually relatively brief and the signs are more marked Cyanosis of moderate or marked degree may be present together with proportional clubbing but these may be absent There is a presystolic mesocardial murmur and accompanying thrill the maximum intensity of these signs being a little to the left of the sternu

in the third and fourth spaces To these may be added the signs of passive congestion of the viscera pulsating neck veins and perhaps an enlarged pulsating liver Enlargement of the heart tends to be greater in these cases than in the Ebstein type

There is no certain method of arriving at a diagnosis of the Ebstein anomaly although it may be suspected in the presence of repeated attacks of paroxysmal tachycardia associated with QRS abnormalities in the electrocardiogram

RADIOLOGICAL PICTURE There is no characteristic picture so far defined of the Ebstein anomaly The heart is enlarged to right and left and there is considerable enlargement of the right auricle In some cases there is massive enlargement of the heart as in the case of Abbott (1936)

ELECTROCARDIOGRAM Few electrocardiograms of Ebstein s anomaly are extant Right axis deviation is usually present and widening of the QRS common There may be right bundle branch block and arrythmias are common

COURSE AND PROGNOSIS In Ebstein s disease the prognosis is better than in the other types Cyanosis may be late in appearance and the condition is not incompatible with long life The average age at death is about 25 years but the age of 60 has been attained in at least two instances Death often results from pulmonary tuberculosis but may be from congestive failure In other types symptoms and cyanosis are early and severe and life is not more than a few years

CHAPTER XV

TRILOCULATE AND BILOCULATE HEARTS

True examples of these types of congenital heart disease depend upon the failure of development of the cardiac septa and are rare Most cases are anatomically four chambered but functionally three or two chambered The cor biatriatum triloculare has recently become important because certain types are amenable to surgical treatment

Cor Biventriculare Triloculare

(Complete absence of the Interauricular Septum)

The defect arises as a result of failure of development of the auricular septum and is often associated with defects of the pulmonary veins or vena cavae as in the case of Davidson (1930) In William s (1894) case the left pulmonary vein entered the superior vena cava and the right pulmonary vein was rudimentary Dextrocardia and transposition of the vessels was present in the cases of Davidson (1930) and Ratner and Abbott (1921) An interventricular septal defect was additionally present in the cases of Davidson Lereboullet (1931) and Yater Leaman and Cornell (1934) which latter also showed congenital heart block The case of Zadoc Kahn and Cousins (1925) was complicated by an aneurysm of the interventricular septum It is a feature of practically all of the reported cases that there is some minute remnant of the auricular septum but functionally it is virtually absent

Clinical Picture Cyanosis may be late in appearance as in the less severe types of septal defect because the shunt is arteriovenous and reversal of flow is only likely to be transient as with pulmonary infection or terminal Some cases have been reported with both cyanosis and clubbing and this suggests the presence of other abnormalities There are no characteristic physical signs and a murmur if present is due to some associated defect The X ray picture may show evidence of right ventricular hypertrophy and the electrocardiogram a right axis

Course and Prognosis Death usually results from cardiac failure although an infective endocarditis claimed the case of Lereboullet Davidson s case died of congestive failure at the age of 27 Abbott (1936) gives the average age at death as 6 with a maximum of 31

Cor Biatriatum Triloculare

A heart consisting of two auricles and a ventricle is designated as a cor biatriatum triloculare In a true example of this anomaly there is

a complete arrest of development of the interventricular septum and both auricles open into a common ventricle from which arises the aorta or pulmonary artery or both in normal or more frequently transposed relationships An example is the case of McCrea (1927) a cyanotic child of 10 weeks in whom there was a single ventricle transposed great vessels and a patent foramen ovale Such cases mostly die early in childhood but a few attain early adult life Again in nearly all the reported cases there is some vestige of the interventricular septum especially its posterior portion

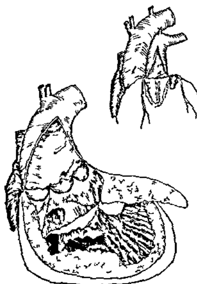


Fig 86 Heart from a child who survived for a few hours An anomalous ventricular septum cuts off a small anterior ventricle from which arises the pulmonary artery Two auriculoventricular orifices open into the large posterior ventricle This heart may be classified as a type of cor biatriatum triloculare

The most important type because it may be clinically recognizable and in some cases amenable to surgery is the single ventricle with a rudimentary outlet chamber in one or other of its variable forms The common denominator of this group of cases is a single ventricle into which both auriculoventricular ostia open and a rudimentary outlet chamber separated from it by a ridge of muscular tissue The aorta or pulmonary artery or both vessels may arise from the rudimentary chamber in normal or transposed relationships

In the development of the heart the ridge intervening between the bulbus cordis and common ventricle disappears Coincidently with this the interventricular septum begins to develop and partitioning of

the truncus occurs Ordinarily only the anterior part of the bulbus persists as the pulmonary conus but in this abnormality the bulbus cordis persists and is represented by the rudimentary cavity At the same time abnormalities occur in the development of the interventricular septum and aortic septum

ANATOMICAL TYPES OR COR BIATRIATUM TRILOCULARE

It is possible to identify three main groups of cases

(1) *Single ventricle with rudimentary outlet chamber from which arises the aorta and pulmonary artery*

Good examples are the two cases of Taussig (1939) The first an infant of four weeks with persistent cyanosis emphasized by crying The right auricle was greatly enlarged A deformed tricuspid valve and a mitral valve opened into a large single ventricle At the site of the pulmonary conus was a rudimentary chamber separated from the large ventricle by a muscular ridge This chamber gave rise to a large aorta The pulmonary artery was obliterated at its base and the ductus arteriosus was widely patent In the second case there were attacks of cyanosis The heart was slightly enlarged There was a gross defect of the auricular septum so that there was virtually a common auricle divided by only a strand of tissue The two auriculoventricular valves opened into a single ventricle A rudimentary cavity existed at the site of the pulmonary conus and was separated from the common ventricle by a muscular ridge The aorta and pulmonary artery arose in trans posed relationship from this diminutive chamber The ductus was closed

(2) *Single ventricle receiving both auriculoventricular orifices Aorta from single ventricle and pulmonary artery from the rudimentary chamber*

These cases are rare (fig 86) The first case was described by Holmes (1824) in a male of 22 and remained unique until the case of Drey Strauss and Gray (1938) This latter case a girl of 14 presented cyanosis and clubbing and died of a cerebral abscess The right auricle was dilated and hypertrophied and both mitral and tricuspid valves entered a single ventricle The aorta arose from the posterior part of this single ventricle and was somewhat narrow in calibre A rudimentary cavity in the left anterior wall of the single ventricle gave origin to a pulmonary artery and communicated by a small opening with the larger ventricle In a personal case a cyanotic female child of 10 presented a left axis in her electrocardiogram and an X ray picture which closely resembled that of the tetralogy (fig 87) An anastomotic operation was performed and the child died post operatively The post mortem disclosed a common ventricle into which both auriculoventricular orifices opened There was a diminutive chamber communicating with the large ventricle by means of a very small interventricular septal defect The pulmonary artery arose from this small chamber

(3) *Single ventricle with rudimentary outlet chamber The great vessels are transposed so that the aorta arises from the rudimentary chamber and the pulmonary artery from the single ventricle*

This type is the commonest and was described by Peacock (1855) in a child of eight months Subsequent cases are those of Rokitansky (1875) associated with pulmonary stenosis Spitzer (1923) two cases one with tricuspid atresia Mills (1923) with coarctation Favorite (1934) Glendy Glendy and White (1944) This does not exhaust all



FIG 87 Single ventricle with diminutive outlet chamber with the great vessels not transposed (Holmes heart) The pulmonary artery arose from the diminutive chamber Marked cyanosis and left axis in the electrocardiogram The case died subsequent to an anastomotic operation

the reported cases many of which have probably been reported under other titles Thus mitral atresia is sometimes described as a triloculate heart In this anomaly the heart may be functionally triloculate and a small hypoplasic left ventricle buried in the musculature of the common ventricle may be missed unless a careful search is made Similarly with tricuspid atresia a hypoplasic right ventricle may be overlooked and it may be assumed that the interventricular septum is absent

Even in those cases where there is a ventricular septal defect identification can be made certain because in the group of anomalies under discussion both auriculoventricular orifices open into the single ventricle and in mitral and tricuspid atresia the corresponding artery is hypoplasic or atresic Favorite s case a boy of 18 presents certain unusual features (fig 89) Both auricles communicated with a large common ventricle the pulmonary artery which was dilated and caused death by rupture arose from the common ventricle A transposed and hypoplasic aorta arose from the rudimentary chamber



FIG 88 Single ventricle with diminutive outlet chamber The great vessels are not transposed A probe is placed in the small foramen communicating between the single ventricle and the small outlet chamber An anastomotic operation has been performed

CLINICAL PICTURE Cyanosis may or may not be present the variation in the picture being due to the situation of the pulmonary artery and the amount of blood that reaches the lungs When the pulmonary artery takes origin from the rudimentary chamber it is small and hypoplasic and cyanosis tends to be severe If however the vessels are transposed

and the pulmonary artery arises from the single ventricle clinical cyanosis is absent or at the most very slight There is however always an increased amount of reduced haemoglobin in the arterial blood Correspondingly an aorta arising from the diminutive chamber implies a reduced systemic blood supply and this is reflected in the frequent under development of these subjects The heart is usually normal in size A systolic murmur may be present and heard widely over the praecordium

RADIOLOGY The conus of the right ventricle is enlarged because of the position of the rudimentary chamber The heart as a whole is not

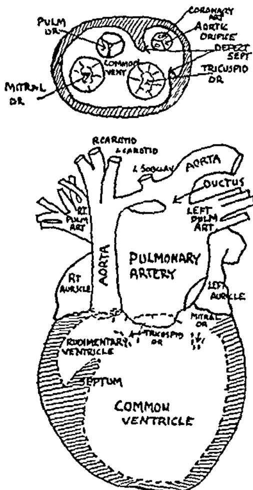


FIG 89 Diagram of cor biloculare (After Favorite)

enlarged If the pulmonary artery arises from the common ventricle there are prominent hilar shadows if it arises from the rudimentary chamber the hilar shadows are minimal In the left anterior oblique position the right ventricle does not project beyond the line of the ascending aorta and the appearance thus differs from that of the tetralogy

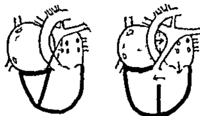


FIG 90 The circulation in tricuspid atresia and in pulmonary and tricuspid atresia

ELECTROCARDIOGRAM Right axis is general but there may be no changes in axis Left axis occurred in a personal case identical with the Holmes heart (fig 91)

DIAGNOSIS This may cause the greatest difficulty but an accurate diagnosis is essential in those cases that are cyanotic so that they can be considered in regard to surgical treatment Surgery can be entertained for those cases where the pulmonary artery is small and arises from the rudimentary chamber Where both vessels arise from the single ventricle even if the pulmonary artery is small the pressure in the aorta and pulmonary artery is likely to be equal and therefore there are no advantages in performing an anastomatic operation because the blood would not course through the channel so created

The silhouette of the heart in the left anterior oblique is not unlike that found in tricuspid atresia and this can usually be differentiated by the left axis in the electrocardiogram In the cases where there is no cyanosis rheumatic heart disease as in the case of Glendy Glendy and White may have to be excluded by the history and symptoms Lastly a sample of blood from the ventricle and a sample of arterial blood should have identical oxygen content if there is a single ventricle

PROGNOSIS Abbott gives the average age at death as 7¾ years in the thirteen cases that she collected

Cor Biloculare

A true cor biloculare in which there has been no attempt at septal formation is exceedingly rare and it is doubtful if it ever occurs in the strictly anatomical sense Keith (1909) and Wood and Williams (1928) have yet to see a true cor biloculare in regard to structure and

they suggest that the term biloculare should refer to function rathe than *structure*

The cor biloculare represents the most primitive type of cardia defect and designates a functionally two chambered heart The reported cases fall into two main groups which may be arbitrarily referred to as complete or incomplete forms according to the exten of septal development *The complete form may be further subdivided* into two classes depending upon whether there is subdivision of the

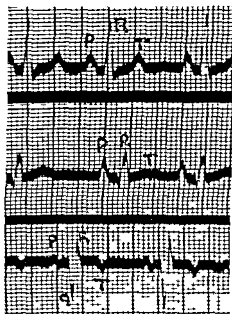


FIG 91 Single ventricle with diminutive outlet chamber Great vessels not transposed Left axis tall pointed P waves deep Q3

truncus arteriosus or whether this latter persists The incomplete forms where there has been considerable septum formation are typified by the persistant ostium atrioventriculare commune

The more complete forms of cor biloculare are rare and generally associated with other grave anomalies such as a persistent truncus arteriosus transposition of the arterial trunks or abnormalities of the pulmonary veins In its complete form there is a persistent truncus arteriosus a single auriculoventricular orifice intervenes between a common auricle and ventricle and four *auriculoventricular cusps are* present these latter representing the original undivided cushions of

the atrial canal Arrest in development takes place before the fourth week that is before the appearance of the cardiac septa In a somewhat more frequent type the truncus has been divided into aorta and pul monary artery In the reported cases these are transposed In Kugel s (1932) case there were four auriculoventricular cusps The pulmonary artery was atresic and posterior to the aorta A right sided aortic arch was present and a patent ductus arteriosus communicated with a right and left pulmonary artery The pulmonary veins entered the

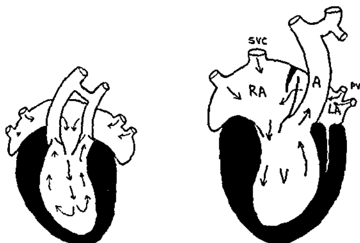


FIG 92 Circulation in the functionally two-chambered heart (After Wood and Williams)

auricle by a common trunk and there was a persistent left superior vena cava A hyperplasic eustachian valve was present in this case as in the cases of Keith (1909) and Wood and Williams (1928) In Michel son s (1943) case an infant of three days there was a large right auricle and a small non functioning left auricle together with a large ventricular chamber Both pulmonary veins entered the right chamber and intervening between the right auricle and ventricle was a poorly differ entiated auriculoventricular valve A single artery emerged from the common ventricle and this vessel was furnished with three cusps Well above the cusps a single vessel was given off which divided into branches entering the lung on either side The main vessel continued as the arch of the aorta with its branches

Transposition of the vessels may be a favourable circumstance and in at least one case that of Rudolf (1899) life was prolonged until the age of 16 In Wood and Williams case (1928) the aorta was dextro posed pulmonary stenosis was present and the ductus arteriosus was patent (fig 92) In Evans s (1933) unique case there was absence of the

ascending aorta The auriculoventricular orifice bore three cusps The pulmonary artery was continuous with the ascending aorta from which the four vessels of the normal arch took origin by a common trunk The number of auriculoventricular cusps is subject to variation and they may be incompletely differentiated three as in the cases of Weiss and Michelson or five as in the case of Wood and Williams Anomalies of the pulmonary veins are of common occurrence Abbott (1927) considered these to be the possible primary abnormality Abnormal pulmonary veins are significant in the genesis of interauricular septal defects Morphologically the partitioning of the auricle depends upon the formation of a pulmonary circulation and disturbances of the blood flow entering the auricle might well lead to absent or anomalous septal formation The inferior vena cava may be absent and in its place hepatic veins may enter the auricle It is evident from the above that the complete forms of the cor biloculare may present a primitive and bizarre picture that it is quite unrecognizable during life and may be the subject of contention after death The point must be made that a biloculate heart is so functionally more often than anatomically

Persistent Ostium Atrioventriculare Commune

In the incomplete type there is evidence of septal formation but defects are found at the base of the interauricular and at the upper part of the interventricular septum The persistent ostium atrioventriculare commune may be taken as the type and this anomaly is of interest because of its frequency in the mongol Indeed of the forty odd cases reported at least half have occurred in the mongolian idiot Embryologically the anterior and posterior endocardial cushions of the atrial canal fuse at an early stage to divide the atrial canal into mitral and tricuspid orifices Subsequently the cushions proliferate upwards and downwards to assist in the closure of the foramen primum and the completion of the interventricular septum Failure of the cushions to fuse and proliferate will result in a common auriculoventricular orifice with defect of the cardiac septa In these cases the auriculoventricular orifice is situated above the interventricular septal defect and it may be furnished with cusps of unequal size of which the anterior and largest of them appears to be formed by fusion of the anterior cusps of the mitral valve with the septal cusp of the tricuspid valve This conjoined cusp runs across the septal defect The papillary muscles in connection with this cusp are found in both ventricles or the chordae may be attached to the upper concave border of the interventricular septum The number of cusps varies but these are usually five of varying size Apart from this general type there may be other associated cardiac anomalies Lightner s (1939) case presented in addition pulmonary atresia and the right pulmonary veins entered the right side of the auricle Goetsch s (1938) case a mongol of 18

months had coarctation of the infantile type a widely patent ductus and bacterial endocarditis of the auriculoventricular valve

Anatomically all the chambers communicate with each other during diastole and then in systole there is intercommunication between the two auricles and the two ventricles Cyanosis is not always in evidence and if present may vary from time to time Marked cyanosis may be a terminal event

There are no characteristic physical signs A murmur has been present in some cases and absent in many The electrocardiogram in the few recorded instances has shown a right axis A large globular heart has occasionally been observed on X ray examination

CHAPTER XVI

DEFECTS OF THE AORTIC SEPTUM

Defects of the aortic septum form a comparatively small but important group Their origin is developmental and the anatomical picture and to some extent the clinical picture depends upon the degree of failure of development of the aortico pulmonary septum Accordingly these cases are classified into the following groups

(1) Complete absence of the aortic septum or persistent truncus arteriosus

(2) Partial defects of the aortic septum leading to a communication between the aorta and pulmonary artery

(3) Aneurysm of the right sinus of Valsalva

PERSISTENT TRUNCUS ARTERIOSUS In the complete form of this rather rare anomaly the aortic septum is absent and a single large trunk arises from the base of the heart partially or wholly from the right ventricle Such cases are rarer than the incomplete forms

PATHOGENESIS As the name implies the defect is due to a failure of the aortico pulmonary septum to develop and consequently the truncus arteriosus persists as a single trunk Humphreys (1932) states that there is always abnormal torsion but no cause and effect relation ship can be established between this and failure of septation As there are four distal bulbar swellings the rudiments of the semilunar cusps and the latter swellings remain undivided there should be ideally four cusps in the common trunk and this is often the case (fig 93) Associ ated failure in development of the sixth arch system leads to the formation of a collateral circulation to the lungs

In other cases there may be an attempt at septal formation the condition then being known as a partial persistent truncus arteriosus If the spurs of the sixth arch are present but fail to fuse the pulmonary arteries arise independently from the common trunk Where there is partial septal formation there may be a short common pulmonary trunk from which the pulmonary branches are given off and occasionally a ductus arteriosus

The earliest case appears to be that of Wilson (1798) a child sur viving for seven days There was a gross defect of the diaphragm so that the heart was in the abdomen The heart itself consisted of a single auricle and ventricle The latter gave rise to a large single vessel which gave off a pulmonary artery and continued as an aorta with its usual

branches The pulmonary vessel divided into right and left branches The pulmonary veins entered the superior vena cava

ANATOMY An anatomical classification has been proposed by Humphreys who recognizes five main types These are as follows

(1) Partial common trunk with ductus arteriosus
(2) Partial common trunk without ductus arteriosus
(3) Complete common trunk with independent origin of pulmonary arteries

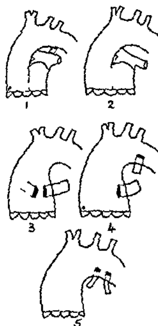


FIG 93 Types of common arterial trunk (Aft Humphreys) 1 Partial common trunk and ductus arteriosus 2 Partial common trunk without ductus arteriosus 3 Complete common trunk with independent origin of the pulmonary arteries 4 Complete common trunk with one pulmonary artery and a bronchial artery 5 Complete common trunk with no sixth arch derivatives and bronchial arteries

(4) Complete common trunk with one pulmonary artery and a bronchial artery
(5) Complete common trunk with no sixth arch derivatives and bronchial arteries

A certain number of cases have been described as a persistent truncus arteriosus when in reality they are cases of pulmonary or aortic atresia (fig 94) Humphreys has suggested certain criteria which may help in the identification of a persistent truncus arteriosus The finding of the

cord like atrophic remnants of either of these vessels and usually an aplastic corresponding ventricle hidden in the wall of a common ventricle should at once *identify the nature of the single trunk* For some authors Hulse (1918) and Humphreys (1932) the presence of four cusps in the single trunk is the hall mark of a persistent truncus and is its infallible sign Hulse would exclude all cases in which three *cusps only were present Humphreys would include those cases in* which there was evidence of a partial division of one of the cusps On

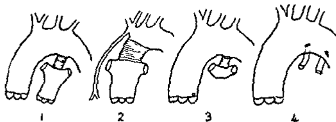


Fig 94 Types of solitary arterial trunk (After Humphreys) 1 Normal separation of aorta and pulmonary artery 2 Atresia of the aorta with solitary pulmonary trunk coronaries arise from atresic trunk 3 Solitary aortic trunk with atresia of the pulmonary artery 4 Solitary aortic trunk with no recognizable sixth arch derivatives Ductus arteriosus shaded

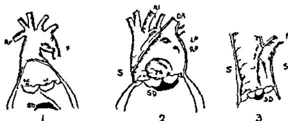


Fig 95 Diagrams of persistent truncus arteriosus (After Feller) 1 Complete persistence of truncus three cusps one with a raphe 2 Per sistent truncus with partial division by an aortico-pulmonary septum Three cusps raphe intervenes between the coronary orifices 3 Partial division of truncus Four semilunar cusps AI aortic isthmus DA ductus *arteriosus* RC and LC *right and left coronaries* RP and LP right and left pulmonary arteries S and S septia SD septal defect

developmental grounds Humphreys would accept cases with three cusps provided *there were no atresic remnants of aorta or pulmonary* artery and the common trunk fulfilled the function of providing blood to both the systemic and pulmonary circulations The exact mode of supply to the lungs depends upon the status of the sixth arch as indi cated above *The pulmonary artery may arise from the common trunk* a short distance above the valves or from the inferior surface of the aortic arch or as in the case of Graham and Montgomery (1938) by a

small vessel distal to the left subclavian artery which divided into branches supplying the left and right lungs Rarely a pulmonary artery has arisen from the descending aorta In other cases the pulmonary blood supply may be through bronchial arteries or from anomalous branches of the innominate and subclavian arteries

A defect of the ventricular septum is inevitable and is usually anterior to the vestiges of the membranous septum (fig 95) There may be almost complete absence of the interventricular septum The inter

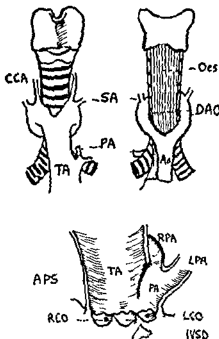


Fig 96 Truncus arteriosus with double aortic arch (After Kerwin)

auricular septum may be patent and a persistent ostium primum has been described There may be a right aortic arch The common trunk arises astride the septal defect or wholly from the right ventricle depending upon the degree of transposition present and in the latter case there is always a septal defect Four cusps were present in the cases of Preisz (1890) Feller (1931) Humphreys (1932) and Brown (1942) The cusps are so arranged that there are two large lateral cusps from the sinuses of which the coronary arteries arise and smaller anterior and posterior cusps Three cusps with evidence of subdivision of one



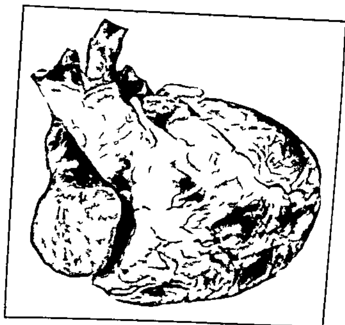


FIG 97 Persistent truncus arteriosus Courtesy of Dr R Marshall

cusp occur in the cases of Buchanan (1864) two cases of Feller (1932) and that of Harris and Thomson (1937) Three cusps only characterize the cases of Zimmerman (1927) Finley (1930) Lev and Saphir (1942) consider the essential features to be the presence of a common vessel from which arises the aorta pulmonary artery and coronary vessels The cusps have been microscopically studied by Roos (1935) and Brown (1942) who showed them to differ from normal cusps by the absence of elastic tissue and their general structure of myxomatous

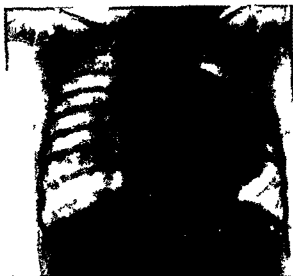


Fig 98 Female child aged 2 years Persistent truncus arteriosus

tissue This finding was not confirmed by Marshall (1943) Enlargement of the heart is generally present A right aortic arch and a double aortic arch have been described (fig 96) In the identical twins reported by Giustra and Tosti (1939) a complete persistent truncus with three cusps was associated with a true cor biloculare Two pulmonary arteries were given off the truncus just above the valve Other examples of a complete truncus arteriosus are those of Graham and Montgomery (1938) and Marshall (1943) In this latter case a large arterial trunk arose from the left side of the aorta about one and a half inches from its origin and divided into a right and left pulmonary artery (fig 97)

It thus appears that there is no constant anatomical picture and the recognition of a case of persistent truncus arteriosus is purely anatomical and depends very largely upon the exclusion of the atresia of one or other great vessel

R

CLINICAL PICTURE Cyanosis may be marked but in some cases it is only slight It depends upon how the pulmonary circulation is secured and is marked when the pulmonary blood supply is derived through bronchial arteries and slight when it is achieved by pulmonary arteries arising from a common trunk The markedly cyanotic case appears to be more frequent Dyspnoea is a prominent feature There are no characteristic physical signs The heart is enlarged to right and left Thrills and murmurs systolic in time have been described over

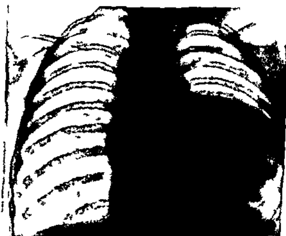


FIG 99 Persistent truncus arteriosus in an infant with a sitting duck silhouette

the base of the heart and a diastolic thrill and murmur was present in the case of Finley and of Pezzi and Agostini A machinery murmur in a lower intercostal space has been described by Crafoord Murmurs are often absent The second sound in the pulmonary area is loud and sharp and very characteristic It is never reduplicated It is so unlike the second sound of other forms of cyanotic congenital heart disease that its presence is very suggestive

RADIOLOGY In infancy there is a cardiac silhouette which is very characteristic of the persistent truncus arteriosus The heart is grossly enlarged to left and right enlargement of the left ventricle pushing the apex upwards There is a marked concavity at the site of the pulmonary arc and the aorta is enlarged with a prominent aortic knuckle The hilar shadows are small and peppered if there is a circulation mainly through bronchial arteries The general contour of the heart resembles a sitting duck (figs 98 and 99) In the left anterior oblique view the appearance is even more striking because the upper margin of the right ventricle projects like a shelf towards the anterior wall of the



(*a*) Posteroanterior



(*b*) Left anterior oblique

FIG 100 Persistent truncus arteriosus Female aged 12 years cyanotic since birth At operation an enormous collateral circulation through bronchial and mediastinal vessels and no visible pulmonary artery

chest With a barium swallow there is a displacement of the aorta backwards towards the spine

In early childhood and later there may be little to distinguish the picture from that of the tetralogy (fig 100 *a* and *b*) This is because the heart assumes a more vertical position with the growth of the child So similar are the two pictures that it may be impossible to be quite sure about the diagnosis without recourse to cardiac catheterization when ventricular blood will have the same oxygen content as blood from the femoral artery

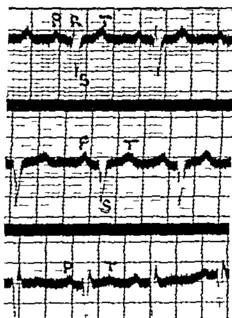


Fig 101 Persistent truncus arteriosus Female aged 14 years Major deflection downwards in all three leads

Electrocardiogram *A few electrocardiograms have shown right* axis deviation In Finley s (1930) case a female of 22 there was a marked left axis Personal cases have shown a marked left axis in one and the main ventricular complex downwards in all three leads in another (fig 101) This latter peculiarity was present in the case of Marshall (1943) (fig 102)

Course and Prognosis The average age at death is given by Abbott (1931) as 4 years The cases of Finley and of Zimmerman (1927) survived to the ages 22 and 25 respectively the former dying of bacterial endocarditis Siegmund s (1928) case died at 33 Death is the

result of failure intercurrent disease or bacterial endocarditis Operations of the Barrett Daley type may be beneficial

Diagnosis *This may be exceedingly difficult in the absence of definite* radiological pointers A catheter is generally freely movable in the ventricular cavity and the pulmonary artery cannot be entered The angiocardiographic picture shows immediate filling of the aorta and in the cases with a bronchial artery circulation delay in filling of the *hilar vessels*

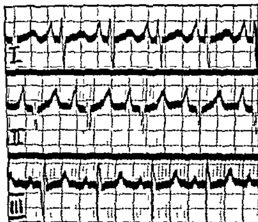


Fig 107 Electrocardiogram in persistent truncus arteriosus The major deflections are downward in the three leads Dr R Marshall s case

Defects of the Aortic Septum and Congenital Aneurysms of the Sinuses of Valsalva

In these conditions there is a communication between the aorta above the aortic valves or at one of their sinuses and either the pulmonary artery or its conus

Pathogenesis Where the communication is between the aorta and pulmonary artery above the level of their cusps there has been a failure in development of the aorticopulmonary septum The communication occurs at sites other than that of the ductus arteriosus Most often the communication is at the base of the aorta *generally in the right aortic* sinus although it may occur in the other two sinuses The opening may communicate with the pulmonary artery above the level of the *pulmonary cusps or it may open into the ventricle at the level of or* just below the pulmonary cusps In the former case the opening corresponds morphologically with the foramen of Panizzae of the crocodile heart where a foramen exists between the contiguous sinuses of the right and left aortae Where there exists a communication with the

pulmonary conus the primary stage of the defect may be a congenital aneurysm of the right aortic sinus which existing initially as a thin walled sac finally ruptures and creates a fistulous opening into the right ventricle The frequency with which an aneurysm of the aortic sinus is associated with a defect of the bulbar part of the ventricular septum suggests that the lesion is due to defective development of the bulbar septum Abbott (1929) refers to a figure of Tandler (1912) in which there is a fusion of the distal bulbar swellings at the level of the



FIG 103 Common aorto pulmonary trunk Antero posterior view (C W C Bain and J Parkinson s case)

cusps thus dividing the aorta from the pulmonary artery com munication existing both above and below the point of fusion

Partial Defects of the Aortic Septum

The defect is a simple circular or oval foraman situated in the anterior wall of the aorta just above the aortic cusps and it leads into the pulmonary artery Such defects arise at about the point where the distal bulbar swellings merge into the aorticopulmonary septum The opening is not in the situation of the ductus and it is not to be con sidered as an atypical or abbreviated ductus

These cases are rare The first reported example may be credited to Elliotson (1830) in whose case there was an aperture admitting a finger where the aorta and pulmonary artery are in contact Wilks (1860) mentions an infant with a similar foramen admitting a goose quill Hektoen (1901) was able to collect nine cases from the literature and

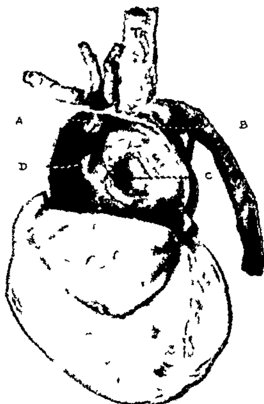


FIG 104 Common aorto-pulmonary trunk Anterior view showing interior of sac A brachio-cephalic trunk B descending thoracic aorta C left pulmonary artery D right pulmonary artery (C W C Bain and J Parkinson s case)

added one of his own Subsequently cases have been reported by Moorhead and Smith (1922) and Bain and Parkinson (1943) The latter case a cyanotic boy of 18 had a defect so large as to con titute an aortopulmonary sac (figs 103 and 104) The case of Dadds and Hoyle (1949) also presented an aneurysmal sac due to a wide opening be tween the aorta and pulmonary artery just above the valves (fig 105)

Shepherd Park and Kitchell (1944) reported a male of 41 who died in congestive failure There was an aperture of 2 cm diameter in the aorta above the cusps This led into a sac which projected into the right ventricle below the pulmonary cusps and in addition the sac communicated with the left anterior sinus of Valsalva of the pulmonary artery by an opening of 1 cm in diameter

Quite often in the partial defect of the aortic septum there is a recognizable ridge on the posterior wall demarcating a segment of the



FIG 105 Congenital aortic septal defect Anterior view showing aneurysmal sac (Dr Clifford Hoyle s case)

common trunk from which one or both pulmonary arteries may arise An example is the case of Kerwin (1936) an infant dying soon after birth and remarkable because of an associated double aortic arch (fig 96) The pulmonary artery arose as a single large trunk just above the sinuses of the left anterior and posterior cusps and promptly divided into branches to the right and left lungs respectively The truncus arose wholly from the right ventricle divided after a course of about 2 cm into two equal sized vessels embracing the trachea and oesophagus to reunite posterior to these structures Pezzi and Agostini (1925) reported a cyanotic girl aged 17 with a diastolic thrill and loud diastolic bruit A common trunk arose from the right ventricle and was

furnished with three cusps A ridge extended from just above the origin of the left coronary artery to the opening of a right and left pulmonary artery Similarly in the case of Brown (1942) there was evidence of a spirally disposed ridge which partially delineated a pulmonary artery

It is therefore evident that an aortic septal defect may exist in its simplest form as a small aperture between the aorta and pulmonary artery and by gradations assume such dimensions as to render its identification from a persistent truncus a difficult matter

The gross pathological result of this particular defect is enlargement of the heart the right ventricle being predominantly involved Dilatation of the pulmonary artery under the influence of a left to right shunt may assume aneurysmal proportions but this is a variable effect depending upon the size of the defect The defect itself has smooth edges in contrast to the more frequent acquired defect where there is always associated aortic disease most often an aneurysm of the ascending aorta Other congenital defects are rarely present Bain and Parkinson s case had a common brachiocephalic trunk This absence of associated congenital defects is of interest in view of the frequent ventricular septal defect associated with congenital aneurysm of the right aortic sinus

Clinical Picture Cyanosis early or late is mentioned in most case reports but clubbing is unusual though noticed by Bain and Parkinson Limitation of activity by dyspnoea or cardiac distress is almost always present Chest pain is frequently mentioned and angina of effort occurred in one case at the age of 18 Congestive heart failure is a common terminal event

The physical signs are variable In some cases only a systolic murmur has been noted at the base In other cases a loud systolic and diastolic murmur is present with or without an accompanying thrill The murmur has been described as continuous more superficial than that of the patent ductus arteriosus and its maximum intensity is lower and more to the right A diastolic murmur and thrill only loudest at the apex was mentioned by Bain and Parkinson whose case failed to show the wide pulse pressure of that of Moorhead and Smith and other authors

Radiological Picture This is very striking in the few reported examples The heart is enlarged chiefly to the right There is an enormous rounded pulmonary arc which obscures the aortic knuckle and renders it invisible The right pulmonary branch may or may not be enlarged A hilar dance has been noted by Dadds and Hoyle (1949) (figs 105 and 106)

Electrocardiogram Right axis deviation has been noted The case of Bain and Parkinson showed a small R and deep S in all three leads

Course and Prognosis The average age at death is about 14 years Rickard s case survived to the age of 31 and that of Moorhead and

Smith died at 48 In these cases the aortic septal defect was small Five of the reported cases died in infancy Most of the cases suffer a severe and *increasing cardiac disability and die in congestive* failure

DIAGNOSIS The striking X ray picture of cardiac enlargement with an enormous pulmonary arc obscuring the aortic knob and pulmonary artery dilatation is very suggestive *The picture must however vary* and where the defect is small the radiological changes may not be so

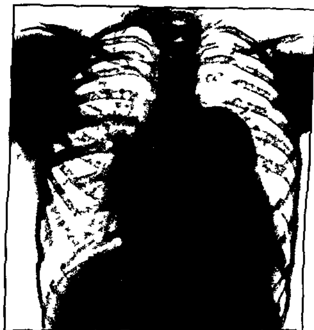


FIG 106 Aortic septal defect Male aged 14 Dr Clifford Hoyle s case

marked The X ray picture may closely resemble that of the auricular septal defect and distinction from this latter anomaly may be very difficult The auricular septal defect is rarely if ever accompanied by the marked physical signs of the aortic septal defect and symptoms are not so severe

In those cases where a systolic and diastolic murmur are present a diagnosis of patent ductus arteriosus may be entertained The pul monary artery in the ductus only rarely assumes the prominence that it does in the aortic septal defect The murmur of aortic septal defect is louder and more superficial than that of the ductus and at a lower site Cyanosis is rare in the patent ductus and only occurs in a few

cases complicated by pulmonary arteriolar sclerosis A right axis in the electrocardiogram is of great rarity in the patent ductus

Congenital Aneurysm of the Sinuses of Valsalva

In this anomaly the communication is between the aorta and the conus of the pulmonary artery and consists usually of a small aneurysm of the right sinus of Valsalva with a perforation at its tip Some confusion exists in the nomenclature of the sinuses of Valsalva The three sinuses are best described as right left and posterior the latter being the non coronary sinus The right sinus is related to the right auricle and ventricle The left sinus is external to the left ventricle and of the three sinuses is the most accessible to X ray examination The posterior non coronary sinus is anterior to the right and left auricles These anatomical relationships determine the direction in which an aneurysm will extend Aneurysm of an aortic sinus must not be confused with an aneurysm of the interventricular septum which arises at a quite different situation but equally ruptures into the right auricle or ventricle In about half of the reported cases there is a small defect of the anterior portion of the septum (bulbar septal defect) just below the aneurysm Abbott s (1919) case is a well known example where an aneurysm of the right sinus projected as a funnel shaped tube into the conus of the right ventricle and directly below this there was a septal defect Infective endocarditis to which these cases in general are vulnerable involved the walls and margins of the aneurysm the septal defect and the wall of the pulmonary conus opposite the aneurysm indicating that there was a shunt from left to right In the case of Shepherd Park and Kitchell (1944) a male negro of 41 it seemed that the aneurysm ruptured into the pulmonary artery late in life There was a foramen 2 cm in diameter above the junction of the right coronary and posterior cusps and thus communicated with the pulmonary artery by an opening 1 cm wide in the left sinus of the pulmonary artery The sac of the aneurysm bulged into the right ventricle just below the pulmonary cusps The case of Herson and Symons (1946) (figs 109 and 110) exhibited an aneurysm of the right and posterior sinuses that of the posterior sinus leading into the right auricle just above the septal cusp of the tricuspid valve There was an interventricular septal defect which also led into the tricuspid valve Macleod s (1944) case a man of 54 showed a fistula from the right aortic sinus leading to the right auricle at the point of junction of the anterior and septal cusps of the tricuspid valve Hirschboeck s (1942) case showed an aneurysm originating in the right sinus and extending through the wall of the ventricle into the right auricle at the tricuspid orifice There was a rudimentary posterior aortic cusp

An aneurysm of all three sinuses is a very rare abnormality Only five such cases are known The most recent is that of Micks (1940) in

a male of 25 (fig 107) Autopsy disclosed an enlarged heart Each sinus was expanded to a large aneurysm or pouch The pouch of the left sinus was visible as a swelling of the left of the pulmonary artery and on the left cardiac border and was the largest of the three aneurysms The aneurysms were deep and extended downwards *excavating the* aortic vestibule The patient died of acute heart failure and complete heart block

Congenital aneurysms of the sinuses whether single or multiple



FIG 107 Congenital aneurysm of all three sinuses of Valsalva A part of left auricle B cavity of left sinus cut across C opening of left sinus with orifice of left coronary just above D opening of right sinus with right coronary orifice just above E opening of posterior sinus F left sinus cut across G cavity of left auricle Courtesy of Dr R H Micks

may occur without associated septal defect They may have a certain medico legal importance as they may be associated with sudden and unexpected death as exemplified by the following two personal (un published) cases A large aneurysm of the right sinus of Valsalva which projected into the right auricle and ventricle in a young man of 27 ruptured into the pericardium death occurring during sleep Similarly in a *girl of 15* with congenital aneurysms of all three sinuses un

expected rupture of an aneurysm into the pericardium caused death (fig 108)

CLINICAL PICTURE The shunt through an aortic septal defect or through a ruptured aneurysm of the right sinus is from left to right and cyanosis is therefore absent unless it appears as a terminal event or under other circumstances such as failure or secondary pulmonary vascular changes which might lead to reversal of flow through the shunt



FIG 108 Congenital aneurysm of the sinuses of Valsalva An aneurysm is present at each of the three sinuses Aortic cusps grossly thickened dilatation ascending aorta The left ventricle has been opened posteriorly Female aged 15 Sudden death from rupture into the pericardium

In the aneurysm of an aortic sinus the condition may be entirely latent and symptomless signs and symptoms only appearing when rupture of the aneurysm takes place In Abbott s (1919) case symptoms suddenly appeared after strain and this was thought to correspond to the perforation of a hitherto latent aneurysm A similar eventuality characterizes the cases of Goehring (1920) Hirschboeck (1942) Macleod (1944) and Shepherd *et al* (1944) In some cases there may be the signs of the interventricular septal defect which may accompany an aneurysm An exacerbation of these signs and the development of severe and urgent symptoms such as pain dyspnoea orthopnoea or the appearance of a murmur of different characters than hitherto or of a diastolic murmur are suggestive findings In Herson s (1946) case there was a

FIG 109 Aneurysm of posterior sinus of Valsalva Right auricle and tricuspid valve Arrow points to a ruptured aneurysm Orifice of ventricular septal defect is immediately below the aneurysm (Dr R Herson's case)

FIG 110 Aneurysm of posterior sinus of Valsalva Aortic valve The sinuses from left to right are right posterior and left ventricular septal defect lies below the fused right and posterior cusps (Dr R Herson's case)

rough roaring systolic murmur loudest over the tricuspid area heard as well in the mitral area with a blood pressure of 200/40 Macleod s case presented an aortic systolic thrill and a harsh systolic murmur over the left chest loudest in the aortic area A harsh systolic murmur with apical systolic thrill a parasternal diastolic murmur and a blood pressure of 300-200/0 was present in Hirschboeck s case Shepherd s case showed a continuous roaring murmur over the aortic area with signs of failure A low diastolic pressure reflects the large shunt but it is not always as low as in the cases above Enlargement of the heart is always present after rupture of an aneurysm has occurred

RADIOLOGY Little is so far known of the radiological picture It should be possible to detect an aneurysm of the left sinus and probably of the right The heart is enlarged to the right

ELECTROCARDIOGRAM There is no typical electrocardiographic picture Encroachment of the aneurysm upon the interventricular septum may cause conduction defects Complete heart block occurred in the case of Micks (1940) and Duras (1944) Nodal rhythm and a right axis was present in Herson s case and right axis with T wave changes in the case of Hirschboeck Auricular fibrillation was present in Macleod s case

DIAGNOSIS An aneurysm of an aortic sinus is a difficult diagnosis in the absence of positive radiological evidence or signs of pressure on neighbouring structures It can be suspected when there suddenly appears in a previously healthy case such events as rapid heart failure and the appearance of a systolic and diastolic bruit not previously present

CHAPTER XVII

TRANSPOSITION OF THE GREAT VESSELS AND COMPLETE TRANSPOSITION

In transposition of the arterial trunks the great vessels are altered in their relation to each other or to their respective ventricles Abbott (1927) states that changes take place in the relative position of the vessels and their ventricles whereby the aorta comes to lie in the path of the unaerated blood from the right ventricle The general result is that the aorta comes to lie anteriorly and in relation to the right ventricle

The earliest observation of transposition of the vessels where the aorta arose from both ventricles is probably that of Stenonis (1671) In his case there was a narrow pulmonary artery and a large inter ventricular septal defect Hare lip and cleft palate were associated anomalies A similar case was described by Sandifort (1777) The aorta sprang from both ventricles arteria itaque aorta enascebatur ex ventriculis ambobus et sanquinem ex utroque accepere debebat He also described the narrow pulmonary artery with two cusps and in reality furnished the first accurate account of the anatomy of the tetralogy of Fallot Meckel (1812) noted the resemblance of the mal formed human heart to that of the lower vertebrates and suggested an origin by failure of development at an early stage Kurschner (1837) observed that in transposition of the vessels there was a reduction in or a lack of the normal spiral twist of the pulmonary artery about the aorta Meyer (1857) explained transposition as due to an arrest of *development at a stage before the vessels have assumed their spiral* course Nearly all the earlier observations seem to be concerned with an anatomical combination of pulmonary stenosis and an inter ventricular septal defect Meyer and Peacock (1866) thought that the primary event was an inflammatory pulmonary stenosis at an early stage and that this by causing a raised pressure in the right ventricle forced the developing interventricular septum to the left and thus caused the aorta to emerge wholly or in part from the right ventricle Where pulmonary stenosis was absent Peacock attributed the anomaly to faulty development of the aorta and pulmonary artery from the primitive truncus The production of this form of malformation is to be ascribed to the irregular divison of the arterial trunk so that the branchial arches ordinarily associated with the portion which becomes the aorta are in connection with the pulmonary artery This deviation from the natural development may take place either after the septum

of the ventricle is completed or while the growth of the septum is in progress so that the transposition may involve the ventricle as well as the vessels

Peacock s work was much in advance of his time He preceded Spitzer in his recognition that a septal defect at its usual site corresponded to the foramen between the aortic ventricles of the turtle heart He also stated that the sinus and infundibular portions of the ri ht ventricle were homologous with the right systemic and pulmonary ventricles of the turtle Further he suggested that in cases where both auricles communicated with the left ventricle in certain cases of trans position the infundibular portion of the right ventricle has been cut off by a septum at its point of union with the sinus of the ventricle that is to say there has been a regression of the true interventricular septum

Rokitansky (1875) considered that every type of transposition could be explained on the basis of an abnormal development of the bulbar septum this being the primary event and pulmonary stenosis and an interventricular septal defect were a necessary secondary sequence Deviation of the aortic septum to the left resulted in a dextroposed or riding aorta and at the same time narrowing of the pulmonary artery caused pulmonary stenosis An interventricular septal defect occurred because of failure of the interventricular septum to meet the abnormally placed aortic septum All the other forms of transposition were he stated the result of abnormal rotation of the aortic septum which being unable to meet the interventricular septum resulted in one or both vessels apparently arising from the wrong ventricle This work was a landmark in the history of congenital heart disease and it is all the more remarkable because it allowed Rokitansky to foresee anomalies that he had not already observed Subsequently for some years workers produced variants of Rokitansky s essential thesis Keith s (1909) contribution is important because more recent work tends to bear out his conclusion that abnormal absorption of the bulbus resulted in transposition In transposition the aortic portion of the bulbus expands and the pulmonary portion atrophies and such a process the exact opposite of that which occurs normally results in an aorta anteriorly placed and in the right ventricle Robertson (1913) after studying the comparative anatomy of the bulbus was mainly in accord with Rokitansky s theory She suggested that if the bulbus developed as a straight tube devoid of changes in its middle part there would be no torsion of the aortic septum This would allow the aortic septum to descend and join the proximal bulbar swellings left to left end and right to right end If various positions of the proximal bulbar swellings were possible all the types of transposition could be reproduced

Spitzer (1923) has examined transposition on a phylogenetic basis

As the animal scale is ascended and the lung breathing types appear so also does septation of the heart become more and more developed in accordance with the necessity imposed by a pulmonary circulation of keeping it and the systematic circulation apart Actually the development of a pulmonary circulation furnishes an impetus to development because of forces inherent in the blood stream and is responsible for the formation of the bulboventricular loop torsion and septation

These hydrodynamic factors are a continuous hydraulic pressure the pressure of a continually pulsating flow and the effects of an increasing volume of the circulation The primitive cardiac tube is not only stretched and widened but because it is fixed at both ends it is lengthened and bent to form a loop Further because of the fixation of the tube at both ends torsion to the right occurs at the bulbo ventricular end and detorsion at the opposite venous end This for Spitzer is the critical stage in cardiac development for if torsion did not occur the two circulations would be side by side and there would be a straight septum Spitzer points out that there are increasing degrees of torsion as the animal scale is ascended it being much less in the reptile than in the mammal Peacock Keith and many others have previously remarked the essential similarity of the deformed human heart to the heart of the lower vertebrates

Spitzer observed that the moderator band (trabecula septomarginalis) in the right ventricle was morphologically similar to the muscular ridges derived from the septum aorticopulmonale in the reptilian heart He also indicated that in the human heart especially in childhood there is an area lying between the crista supraventricularis and the anterior tricuspid ledge which represents in the human heart the out flow part of the reptilian right aorta In the human heart the reptilian right aorta is obliterated by the normal clockwise torsion of the primitive cardiac tube As evidence of the normal clockwise torsion he adduces the spiral course of the bulbar cushions and the winding about each other of the aorta and pulmonary artery If however this torsion is deficient or entirely arrested as it may be in grave anomalies the result is that the various parts are brought out of their normal relation ship with consequent ultimate anatomical malformation According to Spitzer incomplete torsion leads to fusion of the primary bulbar septa and a reopening of the right reptilian aorta with obliteration of the left Consequently the aorta in transposition is not a transposed aorta but a reopened right aorta In lesser grades of detorsion there may be partial persistence of both trunks which then fuse and give rise to an abnormally large aorta as in the riding aorta of the tetralogy of Fallot Spitzer explains the presence of a bicuspid pulmonary valve the almost constant concomitant of the tetralogy as due to a persistence of the reptilian state where normally the great trunks are bicuspid

Spitzer s main contributions have been summarized by Liebow and McFarland (1941) as follows

(1) Emphasis on the part played by the advent of a pulmonary circu lation during the transition from an aquatic to a terrestrial habitat in causing torsion
(2) The principle of torsion in septation of the heart so that crossing of the pulmonary and systemic circulations occurs with a mus cular cardiac chamber for each circulation This is more effective than the tandem arrangement in the gill circulation of fishes
(3) The recognition of a rudimentary right aorta homologous to that of reptiles the crista supraventricularis being a guide to the position of this rudiment
(4) Certain septums may be the result of hypertrophy of such structures as the crista supraventricularis tricuspid ledges or bulboatrial ledge with regression of the true interventricular septum owing to haemodynamic changes
(5) Detorsion with reopening of the right reptilian aorta
(6) Establishment of four main types of transposition dependent upon the degree of torsion
(7) *Emphasis on the fact that in no case is there a true transposition* of any great vessel arising from the right side of the heart to the left if the position of the true interventricular septum is used as the plane of reference

Spitzer s theory has found general acceptance but recently it has been criticized by Lev and Saphir (1937) on the grounds that while it explains admirably the anatomical facts it fails to explain adequately just how these changes are brought about The general trend of recent work (Pernkopf and Wirtinger 1935) is to show by studies in com parative anatomy that abnormalities are not so much the result of abnormal torsion of the bulboventricular area but are equally related to abnormal absorption of the bulbus This is in harmony with the conclusion of Keith (1909) though there is a difference in the method of its accomplishment Lev and Saphir have carried the matter further and after a study of their cases of transposition have concluded that an abnormality in the bulbo auricular spur area and thus an abnor mality of bulbar absorption is a fundamental cause of transposition For a fuller discussion of this work and its interpretation the reader is referred to the original paper of Lev and Saphir

CLASSIFICATION OF TRANSPOSITION The earliest classification is that of Rokitansky (1875) He formulates two schemes In Scheme A the interventricular septum unites with the aortic septum so that although *the vessels are altered in their positions as regards each other* they still arise from their respective ventricles There is thus a corrected transposition

In Scheme B the transposed vessels arise from reversed ventricles

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and the transposition is uncorrected The type of this group which is the largest is complete transposition transpositio vera where the aorta arises from the right ventricle and the pulmonary artery from the left These two schemes are subdivided in considerable detail and certain of the possible varieties Rokitansky had never seen

Vierordt (1901) divided the types broadly into complete corrected and partial transposition In the latter both vessels arise from the same ventricle but in reversed relations This classification is convenient and one that is often used

Spitzer divided cases of transposition into five groups These are

TYPE 1 *Dextroposition of the Aorta* The aorta lies astride a high interventricular septal defect and because of fusion of the right and left aortas it is larger than normal There is counter clockwise rotation of the coronary cusps The crista supraventricularis is hypertrophied and in some cases it fuses with the anterior part of the interventricular septum to obliterate the pulmonary conus (pulmonary atresia) The pulmonary valve is bicuspid and the pulmonary artery stenosed or there is atresia of its orifice

TYPE 2 *Simple Transposition* *In Spitzer s view this is not a true* transposition The aorta arises more to the right and the pulmonary artery more to the left Detorsion removes the left aorta from the left ventricle and consequently it disappears The right aorta lying over the right ventricle reopens Both aorta and pulmonary artery arise from the right ventricle The pulmonary artery is bicuspid and stenotic The coronary arteries are rotated more counter clockwise than in Type I The crista supraventricularis is hypertrophied and may have the appearance of an accessory septum Some cases of the tetralogy come into this category and in a few cases there is no pulmonary stenosis (Eisenmenger complex)

TYPE 3 *Crossed Transposition (Complete Transposition of Rokitansky)* The right aorta is opened and the left closed The aorta arises from the right ventricle and the pulmonary artery from the left The vessels appear with the aorta anterior and the pulmonary artery posterior and they run more or less parallel upwards

TYPE 4 *Mixed Transposition* The general type is that of the cor biatriatum triloculare The true interventricular septum is absent and an anomalous septum formed by the crista supraventricularis and tricuspid ledge cuts off a small chamber from the right upper part of the common ventricle From this small chamber arises the right aorta in transposed relationship to the pulmonary artery which arises from the common ventricle Both auriculoventricular orifices open into the common ventricle

TYPE 5 *Inverse forms of the above*

More recently Lev and Saphir have proposed a new classification as follows

(1) Riding aorta with aneurysm of the membranous septum
(2) Riding aorta with an interventricular septal defect
(3) Aorta and pulmonary artery arise from the right ventricle
(4) Complete transposition
(5) Miscellaneous group

A glance at this latter classification will show that all degrees of transposition may be present from the extreme or complete form (transpositio vera) down to the minor degrees of transposition exhibited in an aneurysm of the membranous septum Transposition will usually include the bulk of anomalies associated with an interventricular septal defect where the aorta arises wholly or partially from the right ventricle A truncus arteriosus communis comes into the same category

A number of the types of transposition mentioned above are more fully discussed under their appropriate headings and to these reference should be made

Complete Transposition of the Great Vessels

In complete transposition of the great vessels the aorta arises from the right ventricle and the pulmonary artery from the left ventricle Both aorta and pulmonary artery are normally developed and consequently the clinical picture differs very considerably from those abnormalities where transposition of the vessels is accompanied by pulmonary or aortic stenosis or atresia

The exact embryological cause of transposition of the vessels remains obscure but all investigators explain it as a purely developmental abnormality Keith (1909) considered it to be due to malabsorption of the bulbus Lev and Saphir (1937) postulate that an abnormality in the bulboauricular spur area which allows increased detorsion at the proximal bulbar ostium and so eliminates the necessity for torsion at the distal ostium is the likely explanation The reader is referred to the work of Lev and Saphir and Farber and Hubbard (1939) for a complete discussion of the various theories of transposition

Complete transposition of the vessels is a not uncommon abnormality This is because in foetal life the fact of transposition has little effect upon the foetus and it is only after birth that the abnormality becomes apparent Survival of the child is then dependent upon the persistence of the foetal openings the ductus arteriosus and the foramen ovale and above all upon the presence of an interventricular septal defect

CIRCULATION The circulation in complete transposition is such that the systemic circulation is maintained by the right ventricle and the *pulmonary circulation by the left ventricle* Venous blood enters the right auricle passes into the right ventricle thence into the aorta and systemic circuit The pulmonary veins return blood to the left auricle thence to the left ventricle and this blood is recirculated in the pul

monary system Survival is only possible if there is a means of admixture of blood between the two circuits Accordingly it is imperative that there should be either a patent ductus arteriosus a patent foramen ovale or a defect of the interventricular septum or some combination of these As a result of the shunts through these persistent orifices the heart which is normal in size at birth rapidly increases in size

ANATOMY The heart in complete transposition of the vessels is a striking object yet may easily be overlooked especially as the heart is

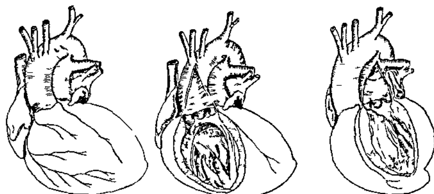


FIG 111 Complete transposition with intact ventricular septum and closed ductus arteriosus Specimen from a stillborn child with spina bifida

small at birth The aorta arises anteriorly and to the right and the pulmonary artery posteriorly The vessels ascend from the heart more or less parallel with each other The coronary openings are situated in the two posterior sinuses of Valsalva and the distribution of the arteries is abnormal (fig 111)

The right ventricle is the same thickness as or thicker than the left ventricle The pars membranacea is muscular There may be a high interventricular septal defect a patent ductus or a patent foramen ovale or auricular septal defect (figs 112 and 113) Where a ventricular septal defect coexists the pulmonary artery may override the defect In those cases where the defect is small or but moderate in size it may be a distinct physiological advantage to the individual In effect it means that venous blood from the right ventricle may be shunted directly into the pulmonary artery and so reach the lungs for oxygenation

In cases which have survived for more than a few weeks the heart becomes enlarged and has a rather globular configuration

CLINICAL FEATURES Cyanosis is the logical outcome of the anatomical abnormality for an aorta arising wholly from the right ventricle passes venous blood into the systemic circulation The only oxygenated blood that can reach the right ventricle is that shunted from the left ventricle through an interventricular septal defect Cyanosis therefore is evident

within a few hours of birth and is rapidly progressive and soon becomes intense It may be mitigated to a slight extent where there is a ventricular septal defect or a widely patent ductus arteriosus In all cases cyanosis early becomes permanent and progressive and if survival is long enough clubbing and polycythaemia develop It is an interesting observation that cyanosis is less marked in the legs in those cases where a patent ductus arteriosus carrying oxygenated blood is continuous with the descending aorta

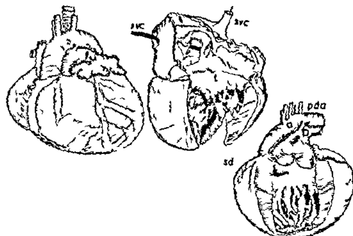


Fig 112 Transposition of the vessels with ventricular septal defect and bicuspid pulmonary valve

The heart is normal in size at birth and if the infant survives it rapidly enlarges the enlargement involving particularly the right side The left side also enlarges if there is a patent ductus arteriosus but not to the extent of the right which latter side carries the burden of the systemic circulation The auscultatory signs are not characteristic There may be a systolic murmur depending upon the associated abnormalities and the pressure changes that they produce in the chambers rather than upon the actual fact of transposition A murmur may not be present at all and murmurs cannot be relied upon to give any diagnostic help

Radiology The radiological picture of complete transposition has been delineated by Taussig (1938) The heart is enlarged to both right and left but right sided enlargement predominates The right auricle is often very distended In the anteroposterior view the shadow normally caused by the pulmonary conus tends to be absent with the result that there tends to be a concavity of the upper left border This is further emphasized by the position of the aorta in front of the

pulmonary artery causing the vascular pedicle to be narrow although this finding may be to some extent obscured by a grossly distended superior vena cava (fig 114) In the left oblique view the aorta and pulmonary artery lie parallel to each other and the vascular pedicle is accordingly widened The origin of the pulmonary artery from the left (systemic) ventricle leads to an appearance of adequate vascularity of the lung fields and later to evidence of congestion in the lung fields

Some difficulty may arise in relation to those cases where a pulmonary



Fig 113 Transposition of the vessels with ventricular septal defect Pulmonary valve bicuspid Marked cyanosis from birth

artery overrides a ventricular septal defect Because in these circumstances the pulmonary artery is of normal size or even slightly larger than normal there may be prominence of the left upper cardiac border and the general appearances may closely resemble those of the Eisenmenger complex Screen examination discloses hilar pulsation Distinction of this type of case will have to be made mainly upon clinical

grounds and it may be taken as essentially correct that whereas in the Eisenmenger complex cyanosis is late in appearance in transposition of the vessels cyanosis is evident at birth and increases with the age of the subject Angiocardiography has been employed successfully by Goodwin Steiner and Wayne (1949) Their pictures clearly demonstrate that the apparently enlarged pulmonary conus in the conventional anteroposterior radiogram was in fact the ascending aorta arising from the right ventricle

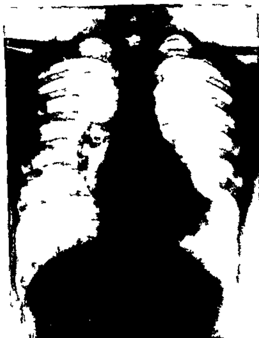


FIG 114 Transposition of the vessels with ventricular septal defect Male aged 20 years

ELECTROCARDIOGRAM The electrocardiogram commonly shows a right axis (fig 115) but occasionally there is sufficient enlargement of the left ventricle to negative this finding A case of this latter type is cited by Schnitker (1940)

DIAGNOSIS Diagnosis is essentially radiological and in the cyanotic case the critical finding is evidence of an enlarged heart an adequate pulmonary circulation and congestive changes in the lung fields Apart from this a narrow vascular pedicle in the posteroanterior view becom

ing wider on rotation to the left oblique view is characteristic In the infant there is usually concavity at the site of the pulmonary arc Diagnosis becomes more difficult in those cases that have survived the first few years of life and who may present a definite prominence of the pulmonary arc and radiological findings not dissimilar to those of the Eisenmenger complex In these circumstances the paramount clinical point is that cyanosis has existed from birth in those cases with trans position whereas in the Eisenmenger case cyanosis develops later A

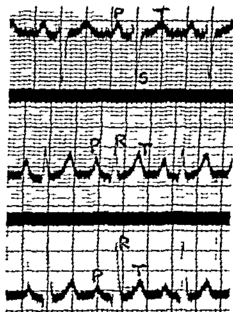


FIG 115 Transposition of the vessels with ventricular septal defect Male aged 13 years Marked right axis Tall P waves

further point of distinction would be that in transposition exercise results in a fall in oxygen consumption whereas in the Eisenmenger there is an increase in oxygen consumption per litre of ventilation

COURSE AND PROGNOSIS In complete transposition unrelieved by a septal defect death usually takes place in the first few days or weeks of life although one case survived to the age of 11 When there is a ventricular septal defect Abbott finds the average age at death to be 2 75 years Exceptional cases of transposition with a ventricular septal defect may live until the second decade or later Survival in these cases may depend upon the position of the pulmonary artery which may

advantageously override a high septal defect or upon the size of the defect itself

TREATMENT Hanlon and Blalock (1948) have begun experiments in the surgery of transposition of the vessels The pulmonary veins are anastomosed to the superior vena cava at the opening of the azygos vein in their experimental animals The successful results of these operations causes the authors to conclude that the operation might be feasible in man and offer a possible approach to the amelioration of complete transposition of the vessels



FIG 116 Corrected transposition Anterior ventricle opened Bicuspid auriculoventricular valve

Corrected Transposition

In corrected transposition the vessels are transposed the aorta lying in front and pulmonary artery behind but in spite of this transposition they are placed in their respective ventricles There is generally an interventricular septal defect and the mitral and tricuspid valves are transposed The anatomical condition present is well illustrated by an unpublished case of the author (figs 116 and 117) In a female child of 10 months with Little s disease the heart was enlarged The aorta lay in front and the pulmonary artery behind The aorta opened into a small anterior ventricle which communicated with the left auricle by a valve with two cusps The pulmonary artery opened into a large posterior ventricle which formed the whole apex of the heart and which communicated with the right auricle by a tricuspid orifice The ductus arteriosus was closed and slight coarctation was present Just below the aortic valves there was a round defect of the ventricular septum The left auricle received two right and one left pulmonary veins The right auricle received a normal superior and inferior vena cava and an additional left superior vena cava which descended to the left of the aorta and ran along the lower margin of the left auricle in the auriculoventricular groove to reach the right auricle The foramen ovale was closed A similar case is described and figured by Theremin (1895) and

also by Rokitansky More recently cases have been described by Stejfa (1931) and Carns Ritchie and Musser (1941)

Not the least interesting feature of several of the reported cases is that the correction of the transposition by the interventricular septum is apparently negatived by the transposition of the auriculoventricular valves for a tricuspid valve is the hall mark of a venous ventricle as is the mitral valve of an arterial ventricle No adequate explanation of this is as yet forthcoming and it cannot be explained by Spitzer s

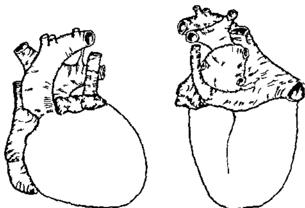


Fig 117 Corrected transposition Female aged 10 months Anterior and posterior views The dotted line indicates the limits of the anterior arterial ventricle

theory Rokitansky conceived eight possible variations of corrected transposition but Spitzer avers that he has never seen an example

CLINICAL PICTURE There are no definite features to distinguish these cases during life and diagnosis is usually made at autopsy In the author s case there were no symptoms or physical signs In Stejfa s case *which attained early adult life* there was dyspnoea on exertion cyanosis and enlargement of the heart A loud systolic murmur was heard over the heart and was loudest in the second left space

RADIOLOGY In the author s case there was some general enlargement of the heart and the vascular pedicle appeared to be small In Stejfa s case there was no enlargement of the heart but there was a large pulmonary arc

ELECTROCARDIOGRAM Left bundle branch block was present in Stejfa s case Complete heart block and a left axis was present in the case of Abbott (1936)

CHAPTER XVIII

DEXTROCARDIA

Dextrocardia is the term applied to those cases in which the heart assumes a position in the right side of the chest with its apex pointing to the right The designation dextrocardia is properly reserved for those cases where the position of the heart is the result of a developmental abnormality and not due to disease or other changes in the adjacent structures

Dextrocardia was first noted by Severinus (1643) in complete situs inversus and is thus one of the earliest recognized heart abnormalities Senac (1749) identified acquired and congenital types Bouilland (1835) differentiated between transposition and displacement of the heart and first recognized isolated dextrocardia clinically

The cause of dextrocardia is obscure and must be sought in the events of the tenth to fifteenth days of intrauterine life in cases where the heart cavities are inverted At this time the heart assumes the appearance of an S shaped tube A kinking of this primitive tube in an opposite direction to normal has been thought to be the cause of dextrocardia

Several varieties of dextrocardia are recognized (fig 118) and they may be classified as follows

(1) DEXTROCARDIA WITH SITUS INVERSUS TOTALIS (Synonyms—Transposition of the viscera complete heterotaxy mirror image dextrocardia) The heart and all the thoracic and abdominal viscera are completely transposed but as their relationship to each other is unaltered a *mirror image* of the normal results

Situs inversus totalis is the commonest variety of dextrocardia and is of no clinical significance Macera and Bordato (1930) were able to collect 250 cases from the literature and since then a considerable number have been added Kartagener (1933) stressed the frequency of bronchiectasis in this condition The discovery of dextrocardia is often accidental at a life assurance or mass X ray examination A familial incidence has been recorded in four generations by Lancisi The genetics of transposition of the viscera have been discussed by Cockayne (1937) There is left handedness in about 40 *per cent* of cases In this the commonest type of dextrocardia the heart is usually exempt from other abnormality The apex beat is in the fifth right space otherwise the heart appears normal and there are no symptoms

Radiography shows a mirror image of the heart and thorax the

aortic arch lying on the right side and the left diaphragm being higher than the right. Barium examination shows the stomach on the right side and the caecum on the left.

The electrocardiogram in the uncomplicated case is pathognomonic. All waves are inverted in lead 1 and leads 2 and 3 change places (fig. 119). Modifications of the electrocardiogram may be found in cases complicated by other congenital cardiac defects. The ventricular complex may be upright in lead 1 as in the cases of Abbott and Moffatt

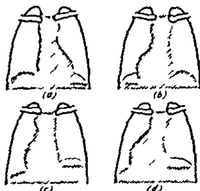


FIG. 118. Diagrams of radiographs in dextrocardia. (a) Normal. (b) dextrocardia with transposition of viscera. (c) isolated dextrocardia with inversion of heart cavities. (d) isolated dextrocardia without inversion of heart cavities. (After Mandelstam and Reinberg)

(1929), Banerjea (1935) and Beaujeu and Benmussa (1939) despite lesions of the Fallot type tending towards right predominance. In other complex cases the P wave may be upright in lead 1 as in the case of Beaujeu and Bege (1937). Cases with coronary disease are those of Manchester and White (1938), Crawford and Warren (1939) and Cain (1945). There are no symptoms in the uncomplicated case. The condition requires no treatment and is entirely compatible with a normal life.

(2) ISOLATED DEXTROCARDIA WITHOUT TRANSPOSITION OF THE VISCERA. In this condition there is heterotaxia of the heart alone with normal position of the viscera. The subject has been exhaustively studied by Lichtman (1931). Other associated congenital anomalies of the heart are frequently described in this group and reference is often made to complicated or uncomplicated isolated dextrocardia depending upon the presence or absence of such abnormalities. Roesler (1930) maintained that every case of isolated dextrocardia was complicated by other cardiac anomaly. Lichtman's study of 161 cases from the literature revealed only three cases in which there was no associated

cardiac malformation Stevenson (1937) published a case of uncomplicated isolated dextrocardia and discussed the anatomical specimen of the same condition prepared by Thomson in 1854 A recent case is that of Beaujeu and Benmussa (1939)

Two main types are described depending upon the presence or absence of inversion of the heart cavities

(*a*) *Heart Cavities Inverted* In this type the heart is transposed in the same way as in complete transposition the right ventricle lying in

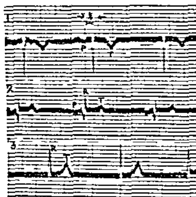


FIG 119 The electrocardiogram in mirror image dextrocardia

front and the left ventricle behind forming the apex The aortic arch may be either on the right or the left side the latter being rather commoner The genesis of the condition is apparently the same as in the situs inversus totalis with the exception that only the heart is involved With the X ray there may be either a true mirror image or the aorta may be on the opposite side Signs of congenital heart disease often of the cyanotic type are frequently present although the condition may be uncomplicated as in the cases of Stevenson and Thomson above

The electrocardiogram may or may not show the alterations characteristic of dextrocardia of the mirror image type The uncomplicated type presents the classical picture of dextrocardia Associated cardiac abnormalities may cause alterations in the picture

The symptoms and signs are those of the associated heart lesion Severe malformations lead to early death while those of the uncomplicated type may attain adult life

(*b*) *Heart Cavities not Inverted* (*Dextroversio Cordis*) In this type which furnishes the larger number of cases of isolated dextrocardia the heart lies on the right but the heart chambers are not inverted

There is thus no mirror image of the normal The aortic arch is usually on the left side The anomaly may arise in the fifth or sixth week of intra uterine life when an arrest of development takes place and appears to be independent of any change in the direction of the primitive S shaped heart It seems to be rather a persistence of the early foetal position of the heart For some reason as yet unexplained the heart appears to have been rotated round its vertical axis from left to right The left ventricle becomes anterior and the right posterior



FIG 120 Apparent dextrocardia due to eventration of the diaphragm The electrocardiogram was physiological

forming the apex *The aortic arch is generally on the left side and the* aorta descends on the left In many of the reported cases pulmonary stenosis or atresia and transposition of the great vessels has been present and the aorta lies in front of and to the left of the pulmonary artery Such changes might be anticipated as the condition arises before the septa have closed The general anatomical picture conforms to a type of corrected transposition of Rokitansky where although the vessels are transposed they yet arise from their respective ventricles because of these anomalies cyanosis is often present In the case of Mayer (1937) a man of 60 with hypertension and isolated dextrocardia the electrocardiogram was normal and diagnosis was made on radiological grounds a transposition of the great vessels being recognized by an anomalous appearance of the ascending aorta in the left anterior oblique position

In cases where an electrocardiogram has been done (Vaquez and Donzelot 1920 Molari 1936 Meyer 1937) there was no change in lead 1

In general the prognosis is bad in this group because of the complex associated abnormalities usually present

(3) DEXTROCARDIA ASSOCIATED WITH CONGENITAL DISEASE OF THE DIAPHRAGM In diaphragmatic hernia or in eventration of the diaphragm the heart is displaced to the right (fig 120) The heart is normal and there are no electrocardiographic changes

CHAPTER XIX

INFECTIVE ENDOCARDITIS IN CONGENITAL HEART DISEASE

The abnormal tissues of the congenital heart defect even of the slightest type are peculiarly vulnerable to infective processes This liability of abnormal tissues and structures to bacterial invasion was first pointed out by Paget (1844) was confirmed by Osler (1866) and Horder (1909) and again emphasized by Abbott (1927) Infective endocarditis is the most serious risk to which the case of congenital heart disease is exposed The actual incidence of infective endocarditis varies with the different abnormalities and is greatest in those cases where there are vigorous shunts or obstructive valvular lesions Thus the highest incidence is in the acyanotic group of cases notably the patent ductus arteriosus interventricular septal defect aortic and subaortic stenosis and bicuspid aortic valves Of Abbott s (1927) 395 acyanotic cases 22 per cent had bacterial endocarditis In contrast the case with the interauricular septal defect of significance even when associated with mitral stenosis so rarely develops infective endocarditis that its occurrence is a pathological curiosity On the other hand the inter auricular septal defect presents a peculiar frequency of associated rheumatic valvular disease most often of the mitral valve This was first stressed by Roesler (1934) and again emphasized by Gelfman and Levine (1942) and subsequently by Burrett and White (1945)

The very interesting analysis of 453 autopsied cases of congenital heart by Gelfman and Levine discloses that the incidence of bacterial endocarditis was 6 5 per cent If however the 181 cases above the age of 2 years are considered the incidence is 16 5 per cent In regard to incidence in specific lesions in cases over 2 years of age their figures are as follows

Auricular septal defects none interventricular septal defects 57 per cent of the uncomplicated cases patent ductus arteriosus 20 per cent bicuspid aortic valves 21 per cent tetralogy of Fallot 29 per cent pulmonary stenosis 29 per cent These figures are perhaps surprising as most have agreed hitherto that the patent ductus exhibits the greatest susceptibility to infective endocarditis (Libman 1910 Abbott 1927) Of equal interest is their finding that 14 per cent of the cases above the age of 2 were complicated by rheumatic disease and bicuspid aortic valves and interauricular septal defects were the most frequent lesions with which rheumatic valvular disease was associated Bacterial endocarditis was present in a third of the cases complicated by rheumatism

Burrett and White (1945) state that 62 per cent of their collected autopsied cases of interauricular septal defect show mitral stenosis although its clinical recognition during life is much less frequent The few cases in which bacterial endocarditis has been associated with an interauricular septal defect are discussed under the latter lesion

In the group of grave anomalies associated with cyanosis infective endocarditis is relatively rare and occurred in 14 per cent of Abbott s (1927) 291 cases and in 29 per cent of Gelfman and Levine s 131 cases The different incidence as between the acyanotic and cyanotic groups may perhaps be due to the greater longevity of the acyanotic case which therefore has an increased risk of infection

The initial site of infection is at some point of mechanical strain This is well exemplified in the patent ductus arteriosus where the infective process begins at the pulmonary end of the ductus and on the wall of the pulmonary artery opposite the ductus opening At this latter site the impinging stream leads to the formation of local ather matous lesions the surfaces of which become covered with platelets and are in their turn infected In the case of coarctation the lesions most often appear either near the site of coarctation or on the ascend ing aorta In the interventricular septal defect vegetations occur on the margins of the defect or on the area of the right ventricular wall where the anomalous shunt impinges In the cyanotic case the chosen sites are the margins of the septal defects or the deformed cusps of pulmonary stenosis The overriding aorta furnishes a direct route for cerebral embolism and cerebral abscess may occur in these cases but only rarely cerebral abscess being more common in the absence of infective endocarditis

The mode of infection is not embolic Damage is done at the points of strain and sclerotic and degenerative changes take place These areas becoming covered with platelets afford a nidus in which organisms circulating in the blood stream may settle and proliferate An inflam matory process is set up and vegetations of remarkable luxuriance appear the le ions spreading but at the same time showing a tendency to heal This mode of origin by direct invasion of the intima was first shown by Hamilton and Abbott (1914) The precipitating event may be a tooth extraction or tonsillectomy

By far the commonest infecting organism is the *Streptococcus viridans* Other organisms are the *B influen a* haemolytic streptococci pneumo cocci and the gonococcus The *Streptococcus viridans* gives rise to an endocarditis of the subacute or Libman type the other cocci tending to cause a more acute and rapidly fatal form of endocarditis in which the endocarditis is merely an incident in a grave septicaemia For some unexplained reason the acute forms often attack the right side of the heart and patent ductus with some frequency (Abbott) The gonococcus has a predilection for the tricuspid valve

The vegetations spring from the affected sites and are often small in the acute case where there may be considerable ulceration of the tissues In chronic cases the vegetations may show some evidence of healing and fibrosis and even calcification Amongst the results of this process are the formation of mycotic aneurysms of the valve cusps or of the membranous septum rupture of the chordae and direct spread to adjacent structures In addition there may be patches of vegetation on the walls of the heart opposite to and in line of exit of an abnormal shunt through a septal defect These are caused by the constant bombardment of the walls by a stream carrying infective particles Such patches may be found on the wall of the right ventricle or on the septal cusp of the tricuspid valve in the maladie de Roger This rather interesting finding recalls the mural vegetations in the left auricle which are a feature of mitral stenosis and which were considered by Thayer (1926) to be the hall mark of pre existing rheumatic heart disease and the mirror of damage previously done to the auricular wall The vegetations consist microscopically of large thrombi formed of fibrin and platelets On the surface of this thrombotic mass are numbers of organisms whilst in the depths of the thrombus there may be numerous leucocytes and red cells The causal organisms appear to flourish on the surface but perish in its depths

From these vegetative lesions emboli may be detached and give rise to the characteristic mycotic aneurysms These result from weakening of the vessel wall by an embolus of a vasa vasorum or weakening of the vessel wall proximal to an embolus They occur most frequently at the bifurcation of vessels lying in soft tissues and are found in the brain heart and peripheral vessels

The types of renal lesion in infective endocarditis are well known There may be infarcts due to embolism of branches of the renal artery The commoner and perhaps clinically the more important type is an embolic focal nephritis due to the arrest in the glomeruli of clumps of streptococci or other organism This occurred in sixty six of sixty eight cases of Baehr (1912) and results in haematuria which may require the microscope for its identification In other cases there may be a diffuse acute glomerulo nephritis

Clinical Features The signs and symptoms of infective endocarditis depend largely upon such factors as the virulence of the infecting organism and the susceptibility of the patient A virulent infection in a susceptible patient may result in a rapidly fatal septicaemia in which cardiac involvement may not be recognized during life In the commoner subacute or chronic types the cardiac signs and clinical features provoked by toxaemia and embolism usually provide a clear cut picture Most often the disease is insidious in onset and the first symptoms so vague that the underlying endocarditis may be unsuspected Common early symptoms are those of lassitude anorexia and low grade fever

Occasionally a catastrophe such as a cerebral or other embolus may lead to recognition of the disease

Once the disease is established there are an infinite variety of symptoms and signs Fever is a constant feature although there may be long periods of apyrexia Apyrexia may be associated with uraemia Rigors are not constant but they usually occur at some period of the disease and are associated with high temperatures and severe sweating Nausea and vomiting may be present so that the patient loses weight and in time becomes emaciated Pain may be present and associated with embolization of various organs or vessels The sternum is often tender Precordial pain radiating to the neck and arms may occur in patients with aortic valve infection Joint pains are common and generally only transient Occasionally the joints are swollen and tender and lead to a diagnosis of rheumatic polyarthritis until emboli or other incidents make the diagnosis clear Pain in other bones may be due to periostitis Horder (1920) refers to the spes endocarditica in which the patient remains optimistic and fails to realize the gravity of his situation a condition comparable to the spes phthisica

There are a whole group of special symptoms and signs of diagnostic importance The spleen is tender and enlarged and infarction of the spleen may be accompanied by severe left sided abdominal pain and tenderness

The cardiac signs are those of the underlying valvular or other structural cardiac abnormality Enlargement of the heart is common Murmurs may change in character or new murmurs appear during the course of the disease Thus the appearance of a diastolic murmur in a case of coarctation may be due to infection of the aortic valve Equally the murmurs may not alter significantly during the whole course of the disease When failure takes place it is generally with a normal rhythm and auricular fibrillation is extremely rare Conduction disturbances are unusual but do occur Libman (1913) remarked on a characteristic pallor the cafe au lait colour of the skin which developed in the more chronic cases and affected the whole body but predominantly the exposed parts Skin lesions may be present and a number of types have been described There may be a rash on the face of butterfly wing distribution resembling the rash of lupus erythematosus and lesions have been described on the wrists and knees A characteristic lesion is the Osler s node present in about half of the cases and first described by Osler (1909) These nodes suddenly appear in the pulp of the fingers or toes as small red painful and tender swellings with white centres and about the size of a lentil They may last for a few days and disappear without trace and are sometimes the first complaint of the patient They are possibly due to small emboli but there is some evidence that they may be a part of a general arterio capillary endothelial proliferation (Jouve 1936) provoked by allergic or toxic changes

in the capillary walls Janeway (1899) refers to small haemorrhages with a slightly nodular character occurring on the palms and soles of the feet The lesions which may be macular or papular are small and erythematous and disappear on pressure leaving a slight brownish pigmentation They may be best recognized if the limbs are elevated for a minute when they stand out in relief against the paler surrounding skin

In the retina haemorrhages may be flame shaped or circular and often have a white centre Boat shaped haemorrhages have been described by Doherty and Trubek (1931) The circular white spots sometimes seen in the retina are referred to as Roth spots or by the French as Litten s sign They are commoner in the more acute forms of bacterial endocarditis

There are other vascular signs which should be sought for Splinter haemorrhages small linear haemorrhages resembling a splinter are found beneath the nails and may be tender on pressure Petechiae the commonest vascular lesion tending to appear in crops or showers may be found in the palms and soles clavicular regions and on the mucous membranes The conjunctiva of the lower lid is a favoured site and the petechiae have a white or yellowish centre and are said to be pathogmonic of the disease In long standing cases the fingers may become clubbed the earliest change being the assumption of a parrot beak appearance of the nails

Ultimately more dramatic symptoms appear and are largely the result of emboli The spleen becomes enlarged and painful and tenderness and pain in the left side may be complained of Hemiplegia may result from embolism of a cerebral artery Cerebral abscess may occur in the cyanotic case Mycotic aneurysms may be recognized in peripheral vessels of the limbs or suspected elsewhere from clinical signs There may be gangrene of the fingers from arterial occlusion The embolic and other lesions of the kidney and their significance have been discussed above It should however be mentioned that some cases die of uraemia Pulmonary embolism occurs when the right side of the heart is involved and in the patent ductus multiple pulmonary infarcts are characteristic

Diagnosis Horder (1926) states that the essential features of the disease necessary for diagnosis are the occurrence of multiple emboli pyrexia and signs of congenital or other valvular disease An enlarged tender spleen and retinal haemorrhages might be added as important corroborating signs A positive blood culture on one or more occasions and isolation of the causative organism is naturally an important help but a negative blood culture does not exclude a diagnosis Cotton (1920) and Merklen (1929) point out the frequency with which petechiae occur Indubitably Osler s nodes and white centred petechiae of the conjunctiva and elsewhere are almost pathognomonic Microscopical

evid nce of haematuria is held to be of great importance and is stated to be present at some time in every case

The protean nature of the signs and symptoms of subacute bacterial endocarditis lead to many complexities in diagnosis The continued wasting and pyrexia may be mistaken for evidence of pulmonary tuber culosis particularly in those cases in which there is recurring pulmon ary infarction as in the infected ductus Rheumatic fever may also be diagnosed in error on the basis of migrating arthritis pyrexia and cardiac signs A positive blood culture or the presence of some typical sign of bacterial endocarditis or the development of nodules or pericarditis will distinguish the two conditions Rheumatic fever and bacterial endocarditis may occur together Typhoid fever malaria and pneumonia can be excluded by appropriate laboratory tests Libman Sacks disease presents the rash of acute lupus erythematosus and is most often in the female Enlargement of the spleen is infrequent Periarteritis nodosa can be recognized by biopsy In sum any acute febrile illness may enter into the differential diagnosis

Course and Prognosis The prognosis once extremely bad is reasonably good with modern therapy It is probable that the spon taneous recovery rate is about 2 per cent Hemsted (1913) reported recovery from an infection involving the right side of the heart Chester (1937) records a case in which a patent ductus arteriosus with subacute endocarditis recovered Other cases exist in the literature but not all of them are convincing The general course of the untreated illness is that of a long febrile disease lasting from four months to two years with steady deterioration of the patient The longer the case lasts the more likely are the major embolic incidents to occur Repeated embolic incidents may occur with periods of apyrexia in which the patient feels relatively comfortable The cause of death may be toxaemia embolism of vital centres uraemia or heart failure

Treatment Horder (1926) strongly advocated treatment along sanatorium lines Until recently there were no specifics and endless remedies have been tried Capps (1922) speaks of cures from the use of sodium cacodylate Att mpts have been made with blood transfusions and mucus transfusion gold salts and other agents For a while the sulphonamide drugs claimed a few successes

The introduction of penicillin has revolutionized the treatment of this hitherto almost untreatable and fatal condition It must however be remembered that however successful the treatment with penicillin may be in abolishing infection a number of deaths are inevitable these result from heart failure uraemia and major emboli

Before treatment is commenced the infecting organism should be identified by a blood culture and its susceptibility to penicillin and sulphonamide drugs tested A bone marrow culture will often give posi tive results when other methods fail (Mallen Hube and Brenes 1947)

Inadequate dosage of penicillin is prejudicial to later success British experience (Christie 1946 Ward *et al* 1946) suggests that the minimum effective dosage of penicillin should be 500 000 to 1 000 000 units a day in divided intramuscular doses three hourly or by a continuous intramuscular drip The dosage is kept up night and day and is continued for twenty eight days Should a relapse occur the course is repeated as many times as appears to be necessary In certain cases a refractory infection may require massive dosage of penicillin combined with sodium *p* aminohippurate (Beyer *et al* 1944 Wood and Felson 1946) This latter drug produces a renal block and leads to a high penicillin level in the blood Recently caronamide has been employed for a similar purpose and results in a fourfold increase of the penicillin level in the blood (Stuart Harris Colquhoun and Brown 1949)

The results of penicillin therapy are extremely satisfactory and many cures have been reported The outlook for these patients is transformed but one must be aware that healing of lesions may increase valvular deformity and lead to failure Penicillin should always be given for prophylactic reasons in patients where tonsillectomy or tooth extraction is necessary

CHAPTER XX

SPECIAL METHODS OF INVESTIGATION

Radiology in Congenital Cardiovascular Disease

Recent advances in cardiovascular surgery have made it important that a correct anatomical diagnosis should be made as often as possible if these benefits are to be applied to the patient A correct diagnosis fortifies any prognosis that may be given It is thus desirable that wherever possible X ray examination should be undertaken in order to acquire evidence of possible anatomical change in the heart and great vessels

The first essential of radiological examination is a standardized method that can be applied to young and old alike For this reason it has been the author s custom to take a photo at 6 ft distance Whilst this method is expensive when compared with an orthodiagram it does not involve the prolonged darkness so terrifying to young children A brief period of darkness is however essential as a prelimin ary to a photograph and the opportunity may then be taken for a fluoroscopic examination Screen examination is of great importance in the observation of the pulmonary arteries and of hilar pulsation The lung fields should not be neglected

The normal radiological anatomy of the heart is depicted in fig 121 which may serve as a guide to the identification of the various regions of the heart The condition of the auricles may be investi_ated in the right oblique position preferably with barium in the oesophagus The left oblique view may be valuable in coarctation of the aorta when it may be possible to identify the stricture of the aorta Recently kymo graphy in the same position has proved valuable in contrasting the pulsation of the aorta above and belo, th- constriction (Laubry and Balsac 1937)

The pulmonary artery is of great importance in congenital cardio vascular pathology and constitutes the second or middle arc of the left cardiac border the lower portion of the curve being formed by the pulmonary conus It is rarely hov e er that these two conponents of the arc can be separated but they may occasionally be recognized in the congenital cyanotic The hila of the lungs consist as shown by Assmann (1928) principally of the division of the right and left pul monary arteries and the accompanying bronchus The left hilum is often hidden by the overlying pulmonary artery and conus The pulmonary artery and conus may be seen in the left oblique positio

running across the space beneath the aorta and just beneath the left bronchus This has been very clearly demonstrated and figured by Schwedel and Epstein (1936) This view admirable as it is in cases where the pulmonary artery is dilated and consequently gives a denser shadow is almost inapplicable in children and has a limited application where there is hypoplasia of the pulmonary artery Bedford and Parkinson (1936) first drew attention to the value of examination of the pulmonary artery with barium filled oesophagus Below the aortic

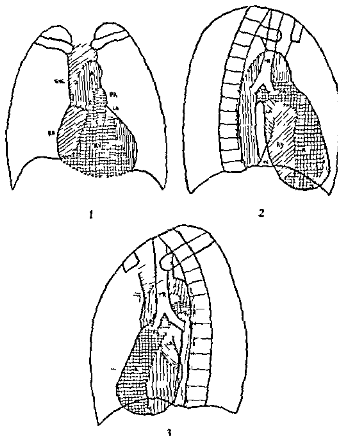


FIG 121 Radiological anatomy of the heart (After Parkinson 1933) 1 Anterior 2 right oblique 3 left oblique position

impression and best seen in the right oblique view is a second impression lying above that of the auricle This impression is most marked in those cases with pulmonary artery disease and enlargement and in these circumstances will be equally obvious in the antero-posterior position of the patient

Opinions have differed widely as to the value of X ray examination in relation to the diagnosis of congenital heart defects All are agreed that in the very young infant with the obvious exceptions of conditions such as idiopathic hypertrophy and situs inversus radiology does not always offer much that is not discernible by the ordinary methods of examination Undoubtedly there are numerous cases where the X ray picture furnishes no assistance in diagnosis in a subject with obvious physical signs On the other hand radiological examination may give confirmatory evidence in a clinically suspected lesion and may in some lesions furnish a diagnostic picture It may also suggest the presence of some other unsuspected anomaly In many cases surviving infancy *that is to say cases with abnormalities compatible with life it is possible* to make a quite reasonable anatomical diagnosis We have recognized dextroposition of the aorta in infancy and have been able to confirm this by serial examination over a period of years with ultimate autopsy Perry (1931) working in school clinics was of the opinion that the X ray examination was at that time of little value Muir and Brown (1935) working in a similar milieu considered that such examination was invaluable Roesler (1937) in a group of sixty eight cases of an average age of 16 8 years found definite positive abnormal radiological findings in 85 per cent these being of diagnostic value in 60 per cent

There must of course be frequent difficulties in interpretation of the radiological picture caused by the presence of combined lesions and by the possible coexistence of acquired valvular disease Such may be the case in the interauricular septal defect The radiological aspects of certain anomalies are at times obvious Dextrocardia is shown by a mirror image picture of the heart and the presence of a situs inversus totalis can be confirmed by a barium meal A right aortic arch can be readily identified

The most important and readily recognizable radiological changes to which attention may be directed are discussed below under their appropriate headings The characteristic pictures presented by certain anomalies are considered under the individual lesions

CHANGES IN THE AORTIC KNOB The aortic knob may be small inconspicuous or even absent in coarctation of the aorta interauricular septal defect and conditions associated with hypoplasia of the aorta Its presence on the right side with displacement of the oesophagus and trachea to the left and forwards characterizes a right aortic arch

CHANGES IN THE PULMONARY ARTERY Changes in the pulmonary artery are among the most striking in congenital cardiovascular roentgenology The pulmonary artery and its conus occupy the middle arc of the left border the hila of the lungs or lung roots being largely made up of the right and left branches Dilatation of the artery or hypertrophy of the conus is shown by a bulging of the arc the part corresponding to the conus being slightly lower than that representing

the artery The right pulmonary branch is clearly visible the left branch is often hidden by the conus and is revealed by rotation of the subject Dilatation of the pulmonary artery occurs in a patent ductus arteriosu aortic septal defect pulmonary stenosis with a closed septum the Eisenmenger complex ind in interauricular septal defects Aneurysmal dilatation of the artery may occur in the latter as also in a patent ductus arteriosus Very rarely an enormous pulmonary artery may accompany an interventricular septal defect Hypertrophy of the

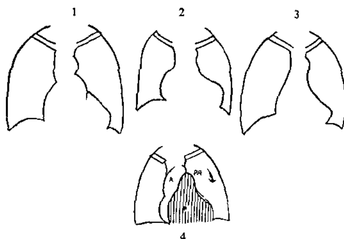


FIG 122 The cocur en sabot 1 Pulmonary stenosis 2 Fallot 3 Fallot 4 to show part played by right ventricle

conus may occur in conus stenosis of the pulmonary artery and in a few cases of the tetralogy of Fallot A rarer cause of dilatation is an infective arteritis Widening and pulsation of both hila is known as Assmann s sign of pulmonary dilatation and is observed in the patent ductus and other conditions of dilatation of the pulmonary artery

The causes of dilatation in valvular stenotic lesions are little under stood unless they are allied to defects in the walls of the vessels Fxperi mental ligature of the vessel in animals has shown a poststenotic dilatation In other conditions dilatation is due to a shunt and to over loading of the right side of the heart

A straight left border to the heart may be found in the maladie de Roger A concave pulmonary arc corresponding to hypoplasia or atresia of the pulmonary artery is found in the tetralogy of Fallot (fig 122) Diminished hilar pulsation and small hilar shadows with marked translucency of the lung fields characterize the tetralogy

ENLARGEMENT OF THE RIGHT VENTRICLE When the right ventricle enlarges the enlargement is almost exclusively to the left of the mid

line With enlargement of the right ventricle there may be an alteration in the contour of the heart giving it a coeur en sabot silhouette (fig 122) The apex of the heart is blunt and raised above the diaphragm the hypertrophied right ventricle being visible as a convexity below the apex A coeur en sabot may occur in pulmonary stenosis with or without an interventricular septal defect and in any condition causing right ventricular hypertrophy The left anterior oblique view furnishes important information

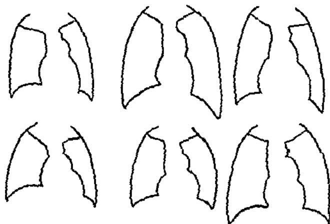


FIG 123 The cardiac silhouette in patent ductus arteriosus with machinery murmur

ENLARGEMENT OF THE LEFT VENTRICLE The chief indications are an increased curvature of the lower arc of the left cardiac border (fig 123) The apex is depressed and displaced to the left The general impression is of hypertrophy of the heart In the left anterior oblique view enlargement of the left ventricle is readily recognized A hypertrophied left ventricle is found in slight degree in subaortic and aortic stenosis in coarctation of the aorta where hypertrophy may be marked if there is aortic incompetence and occasionally in the patent ductus It is a feature of abnormal origin of the left coronary artery from the pulmonary artery

ENLARGEMENT OF THE HEART AS A WHOLE In infants and young children large hearts occur in the so called idiopathic hypertrophy glycogen disease abnormal origin of the coronary arteries aortic atresia mitral atresia tricuspid atresia persistent truncus arteriosus and in the cor triloculare At all ages a large heart is present in the interauricular septal defect and in anomalies complicated by aortic incompetence

ABNORMALITIES WITH A CHARACTERISTIC RADIOLOGICAL PICTURE

These are

- Coarctation of the aorta
- Patent ductus arteriosus
- Interauricular septal defects
- Pulmonary stenosis with closed septum
- Eisenmenger s complex
- Tetralogy of Fallot
- Right aortic arch
- Dextrocardia
- Idiopathic hypertrophy of the heart

These are considered under each lesion separately

The Electrocardiogram in Congenital Heart Disease

Einthoven (1908) first registered an electrocardiogram of congenital heart disease and opened up a new field destined to become an integral part of cardiac examination Steriopulo (1910) and other early workers in the electrocardiography of congenital heart disease assumed that right axis deviation was the general finding in these circumstances Usoff (1911) noted variations in the electrocardiogram of four cases of patent ductus arteriosus and thus destroyed the conception of a specificity of right axis deviation in congenital heart disease

It is perhaps peculiar that there have been so few reviews of the electrocardiogram in congenital heart disease The contributions of McCulloch (1916) and Roesler and Kiss (1931) covered 112 cases but there were only eleven autopsies Similarly the reviews of Battro and Quirno (1935) Katz and Wachtel (1937) and Drawe Hafkesbring and Ashman (1937) lack post mortem control Schnitker (1940) assembled 108 cases from the literature with autopsy control He reached the quite reasonable conclusion that with certain exceptions such as situs inversus there were no features persistently occurring to establish the presence of a specific lesion

At the outset it should be realized that the electrocardiogram is most often physiological and shows but little alteration from the normal It is with rare exceptions in no sense diagnostic It is useful in that it adds to the completeness of the clinical picture and affords general confirmatory evidence

The general features of the normal electrocardiogram of the adult are well known As the investigation of congenital heart disease will mostly be concerned with children the essential features of the electro cardiogram in early age are briefly recapitulated In childhood the P wave in lead 1 tends to be larger than in the adult and in lead 2 may reach a height of 2 5 mm Inverted P waves are not uncommon in lead 3 The PR interval varies to a slight extent with age and up to about the fifth year is about 12 secs At 14 it is said to be about 14 secs

The upper limit of normal is about 18 secs The QRS complex shows a right axis deviation at birth and with the ensuing months as the heart s axis rotates to the left a normal axis deviation becomes evident A low voltage QRS in all leads is rare in childhood The Q wave is large in lead 3 if right axis deviation is present The R wave is normally taller in lead 2 than in lead 1 and an R wave excess of 18 mm may be considered as abnormal Slurring of the R waves if at all marked may be considered as significant The T wave amplitude is about the same as in the adult Inversion of the T wave is common in lead 3 The rate of the heart varies with the age of the patient The approximate rates at various ages are given in the table below

Birth	130-150
1st month	120 140
1 6 months	about 130
6 12 months	about 120
1 2 years	110-120
2 4 years	90 110
6 10 years	90 100
10-14 years	80-90

It has been stated that abnormalities of the electrocardiogram of a degree that would be suggestive in an adult are generally indicative of caridac damage in a child

The only pathognomonic electrocardiogram is that of the situs inversus In this anomaly all the waves of lead 1 are inverted and leads 2 and 3 are transposed (fig 119) There are a few exceptions to this rule as when a case of dextrocardia is complicated by other severe defects or when bundle branch block is present An electrocardiogram will differentiate between a congenital and an acquired dextrocardia there being no inversion of the P wave in lead 1 in the latter condition

One of the principal purposes of an electrocardiogram is to obtain confirmatory evidence which when taken in conjunction with the clinical findings will lend support to an anatomical diagnosis There fore it is evident that the findings of right or left axis deviation may cast valuable light upon the nature of a defect as changes in axis deviation speak for the relative mass of the two ventricles Right axis deviation is found chiefly where there is pulmonary stenosis with or without a ventricular septal defect the most marked degrees being found in the former (fig 124) It may also be found in other conditions tending to right sided hypertrophy and is almost invariable in the auricular septal defect and in tricuspid stenosis and insufficiency A cor pul monale always causes a right axis or a tendency thereto but not necessarily of marked degree It must be remembered that in the early weeks of life there is a right axis deviation so that electrocardiographic examination in extreme infancy may not have the significance and confirmatory value that such an examination may have at a later age

Right axis deviation may occur in lesions of the acyanotic group and then depends upon the position of the heart In the Eisenmenger anomaly it precedes the development of cyanosis In the patent ductus arteriosus where a normal electrocardiogram was considered as characteristic by Lewis (1928) the presence of right axis deviation has been suggested by Laubry and Pezzi (1921) to be evidence of the presence of some complicating abnormality The same reasoning has been applied in the maladie de Roger where a normal axis is the rule

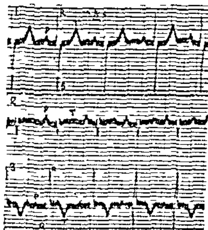


FIG 124 Right axis deviation Tetralogy of Fallot Age 14

The paucity of cases in which an electrocardiogram is supported by autopsy evidence prevents any dogmatic statement relative to these points and the form of the electrocardiogram may not always be consistent with the autopsy findings Repeated electrocardiograms in the same subject are usually consistent with the possible exception of the auricular septal defect It may however be said that right axis deviation found in a case where mitral stenosis kyphoscoliosis and fibrosis of the lungs can be excluded is generally evidence of a congenital etiology the more so if there is a basal systolic thrill and murmur It is probable that right axis deviation is the invariable concomitant of pulmonary stenosis

Left axis deviation (fig 125) may occur in those conditions where there is obstruction in the outflow portion of the heart as in aortic stenosis or coarctation or where there is left ventricular *hypertrophy* from other causes Left axis deviation in the absence of bundle branch block in a cyanotic case at once rules out pulmonary stenosis and is generally due to tricuspid atresia but may occur in a persistent truncus arteriosus and occasionally when there is a common ventricle and right

ventricular hypoplasia Normal axis deviation does not exclude aortic stenosis or coarctation but may suggest the presence of other anomalies when the signs of an obstructive aortic lesion are present In some cases years may elapse before left axis deviation becomes apparent in the electrocardiogram

Abnormally tall ventricular complexes when they occur are suggestive of congenital heart disease A height in excess of 18 mm above the base line may be considered abnormal Such complexes are not so

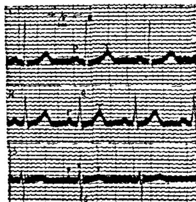


FIG 125 Left axis deviation Subaortic stenosis Age 16

frequent as is commonly supposed and were found in all three leads of 6 to 190 cases examined by Brown and Muir (unpublished) A tall ventricular complex may occur in one lead especially in those cases where there is marked abnormality of axis deviation Perry (1931) suggests that a tall ventricular complex may be a suggestive finding in the absence of any marked ventricular preponderance (fig 126)

Roesler (1931) drew attention to the frequency with which diphasic ventricular complexes were found in congenital heart disease Petit (1926) suggested that a diphasic complex was due to asynchronism of the ventricles the tracing indicating successively predominance of one or other of the ventricles Examination of the tracings show by comparison with the first phase of the diphasic complex in leads 1 and 3 which was the predominating ventricle This ingenious explanation lacks corroboration although it has been suggested that diphasic complexes may be associated with reduplication of the first sound at the apex (Talley and Fowler 1936) Katz and Wachtel (1937) have surveyed 43 congenital cases accepting as a standard diphasic wave one in which the smaller phase has an amplitude of one fourth or more of the larger They found 56 such complexes out of a possible 129 their frequency being 25 times in lead 1 14 in lead 2 and 16 times

in lead 3 They concluded that a diphasic wave of the amplitude indicated was confirmatory in diagnosis and that where the two phases were large and of equal extent they were pathognomonic of a congenital lesion A personal series of 300 cases similarly surveyed showed such complexes in 30 per cent It seems probable that the diphasic wave may have a certain confirmatory value but much remains to be done before its definite acceptance as a pathognomonic feature In the author s experience diphasic complexes are not uncommon in children with rheumatic heart disease

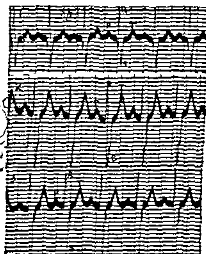


FIG 126 Abnormally tall ventricular complexes in congenital heart disease

An interesting group of cases are those where there is a downwards major deflection of the QRS in all three leads (figs 101 and 102) Such findings have been described in the Eisenmenger complex by Talley and Fowler (1936) and in the patent ductus arteriosus by Schnitker (1940) A personal example occurred in the tetralogy of Fallot and in a persistent truncus arteriosus This finding is not completely explained but it appears to be an expression of right ventricular hypertrophy

Seaham (1924) suggested that prolongation of the ST interval was a characteristic of these cases Our own series does not lend support to this view The T wave may occasionally be negative in lead 2 particularly in cases with right axis deviation The T wave is quite commonly negative in lead 3

An exaggerated P wave occurs in about 20 per cent of cases and is associated with auricular hypertrophy such as occurs in the cyanotic

group or in the interauricular septal defect The P wave is large in lead 2 in these conditions and may be tall and pointed particularly in cyanotic cases with pulmonary stenosis or atresia and interauricular septal defects An inverted P wave in lead 1 is pathognomonic of the situs inversus The P wave may be inverted occasionally in lead 2 and frequently in lead 3 An inverted P wave in lead 3 may be associated with left axis deviation or with hypertension or may occur commonly in the absence of either of these conditions The P wave is inverted and

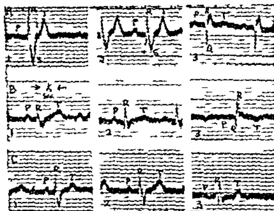


FIG 127 Abnormal electrocardiograms in congenital heart disease

follows the ventricular complex in nodal rhythm as in the case of Laubry Soulie and Vincent (1938) A notched P wave is very occasionally present

The abnormalities of conduction (fig 127) usually found are lengthening of the PR interval auriculoventricular dissociation and bundle branch block Arborization block has been reported by Nissé (1928) The electrocardiogram is the sole method of accurately detecting such changes A lengthening of the PR interval is not so common as might be expected and serial examinations have sometimes suggested that such changes are only temporary and are possibly the result of a rheumatic process which may occur here as in any other heart In two cases a lengthened PR interval observed by the author has been replaced by permanent block In the absence of the rheumatic infection which may be excluded in its active phases by a sedimentation test a raised PR interval is suggestive of an interventricular septal defect when the other clinical findings are appropriate

Bundle branch block has been stated to be only of occasional occurrence in congenital heart disease (Sampson 1948) but personal experience differs from this view The characteristics of typical bundle

branch block are widening of the QRS complex in excess of 01 sec notched QRS either right or left axis deviation depending upon which branch of the bundle is affected and a T wave in opposition to the main ventricular deflection Where sections have been made in bundle branch block it generally transpires that both branches are involved in the pathological process Bundle branch block may also be associated with complete auriculoventricular dissociation and in this case may be attributed to a defective arterial supply because the lower part of



Fig 128 Congenital heart block in a female child aged 3 presenting signs of an interventricular septal defect Gross enlargement of the heart

the common bundle and the first part of the branches are both supplied by the posterior perforating branch of the right coronary artery The rest of the right branch is supplied by branches of the left anterior descending coronary which also supplies the lower part of the left bundle While it will be appreciated that most cases of bundle branch block arise from arterial causes it may also result from developmental abnormalities of the interventricular septum and of the lower part of the auricular septum both of which may entail damage to or deflection of the bundle from its normal course It has been reported in cases of the tetralogy auricular septal defect anomalous coronary artery coarctation and tricuspid valve disease Atypical forms appear to be more common than the well defined right and left types and occur most often with the auricular septal defect These changes may gradually develop over a period of years The prognosis of bundle branch block in congenital heart disease is better than the average but generally poor with the atypical forms Cases which survived more than two years from discovery seem to have an increased expectation of life Complete heart block can only be recognized with certitude by the electrocardiographic picture because complete dissociation may occur with a comparatively fast rate The finding of complete block in an obviously congenital case suggests a defect of the ventricular septum

(figs 57 and 128) Transient heart block has been reported by Wadman (1945)

Nodal rhythm is occasionally present In Calo s (1937) case of the tetralogy the impulse originated alternatively from the sino-auricular and auriculoventricular nodes giving rise in the latter case to the well known characteristic P wave changes Auricular fibrillation is very rare and occurs most frequently in cases of the interauricular septal defect complicated by mitral stenosis or hypertension It also occurs in cases of coarctation Auricular flutter has been present with an interauricular septal defect Electrical alternation of the ventricular complexes is not uncommon and is of undetermined significance

Further detailed surveys of the electrocardiographic picture in congenital heart disease will be found in the works of Perry (1931) Stejfa (1931) Brumlik (1936) Laubry and Soulie (1939) Schnitker (1940) Mannheimer (1940) and Eisenberg and Gibson (1941) To these the reader is referred

In summary it may be said despite the opinion sometimes given to the contrary that the electrocardiogram has a certain value in congenital heart disease as a complement to the clinical examination

Angiocardiography

Forssmann (1931) showed that it was possible to pass a ureteral catheter through the great veins of the neck into the right auricle This observation was the foundation of considerable experimental work notably in Portugal and Latin America whereby radiopaque substances were introduced into the right auricle with a view to outlining the pulmonary tree Ameuille and his co-workers (1936) published photographs of the pulmonary tree obtained by this direct method Castellanos Pereiras and Garcia (1938) succeeded in obtaining excellent pictures which displayed alterations in cardiac topography such as septal defects and pulmonary stenosis These methods have been developed by Robb and Steinberg (1938) Steinberg Grishman and Sussman (1943) Sussman Grishman and Steinberg (1943) and Weber (1943) Its technical value has been assessed by Rayes Castellanos and Pereiras (1943) and by Taylor and McGovern (1943) Angiocardiography calls for a high level of technical skill and special apparatus is essential not only to rapidly introduce the contrast medium but also a rapid plate changer is necessary Preliminary pulmonary sensitisation tests are essential The child is anaesthetised with cyclopropane and liberal oxygen A vein in the antecubital fossa is dissected out and a large cannula inserted A special syringe with a nozzle of wide bore is desirable and 15-20 cc in children and up to 45 cc in adults of contrast medium is then injected within two seconds into the vein Films are taken during the injection and at intervals of 1 to 1½ seconds The patient may if necessary be then placed in the

left anterior oblique position and a further series of films taken In the unanaesthetised patient a sensation of intense heat and often actual pain is produced over the whole body Vomiting may also occur Some workers introduce a catheter into the right auricle through the arm route and use this to introduce the contrast agent Chavez Dorbecker and Celis (1947) pass a catheter into the jugular vein There is accumulating evidence that the procedure is not without its dangers and fatalities are known Perhaps the chief danger of the angiocardiographic examination is that in a very severe cardiac anomaly in which the subject already betrays signs of a dangerously low oxygen saturation the injection of contrast medium may further decrease the supply of oxygenated blood That such dangers exist in relation to pulmonary arteriovenous aneurysms has been shown by Sisson Murphy and Newman (1945) who have produced evidence of leaking of the material in the aneurysms and interference with the oxygen exchange in the lungs In the author s opinion this particular investigation should not be a routine performance on every case of congenital heart disease

For a fuller discussion of the technique and results of angiocardiography the reader should consult original sources and the excellent review of Sussman and Grisham (1947) is recommended Keele (1948) should also be consulted

In the normal case the right auricle is filled in about one second Some contrast medium may linger in the innominate vein The right ventricle is then rapidly filled and it may be possible to identify the tricuspid valve The general appearance is that of a V shaped shadow the limbs being formed by the shadows of the superior vena cava and right auricle on the right side and the right ventricle and pulmonary conus on the left side The pulmonary arteries may also be readily identified in the three second film About six seconds after the injection the left auricle is filled and occupies the gap in the V shadow of the right heart The left ventricle and aorta then fill In the left anterior oblique view it is possible to identify the auricular and ventricular septa These findings will necessarily vary in the case with abnormal anatomical structure of the heart For example concurrent filling of both ventricles may be observed in the interventricular septal defect similarly both aorta and pulmonary artery may be filled at the same time in cases of tetralogy In severe stenosis or atresia there is delay in filling of the pulmonary artery It has been possible to identify the site of a stenosis in pulmonary stenosis and the stenosed segment in coarctation of the aorta Obviously the procedure is a valuable one and it may have important bearings in the pre operative investigation of a case

Cardiac Catheterization This procedure may be of great value in some cases particularly of cyanotic congenital heart disease where the diagnosis remains in doubt and the question of operation is under consideration By this method samples of blood from the various

chambers and vessels may be obtained for analysis of their oxygen or other chemical content pressure measurements may be made at various sites and lastly diodrast may be rapidly injected for X ray photographs aiming at the delineation of the chambers and vessels

For full details of the technique of introduction of the catheter the reader is referred to the works of Cournaud and Ranges (1941) and McMichael Briefly the procedure is as follows

Under local anaesthesia and with full aseptic precautions the median basilic vein at the elbow is exposed and a metre catheter similar to a ureteral catheter with an opening at its distal end is introduced into the vein and passed towards the heart It is necessary to use a fluoro scopic control and the catheter may be manipulated into the right auricle right ventricle and pulmonary artery Where there is an inter ventricular septal defect it may be possible to manœuvre the catheter into the left ventricle and even into the aorta At the various levels pressures are measured by a manometer connected to the catheter and samples of blood are collected under oil At the end of the examination 20 40 ccs of diodrast may be rapidly injected for contrast radiography

It is desirable to have a definite plan in regard to the operation and it is customary for this examination to be performed on patients in hospital Geiger *et al* (1946) state that they have used this method with out patients with no particular preparation and have allowed them to return home immediately afterwards In the very young child it may be necessary to introduce the catheter via the saphenous vein and approach the right auricle through the inferior vena cava

Customarily samples of blood are collected from the superior vena cava the right auricle right ventricle pulmonary artery and a distal branch of the pulmonary artery If the other sites are reached samples should be collected from these as well The oxygen content of these samples is then estimated using a Haldane apparatus

Oxygen estimations furnish information of the greatest value In an interauricular septal defect the oxygen content of right auricular blood is significantly increased in comparison with that of superior vena cava blood because of the arterialized blood shunted through the defect from the left side of the heart For similar reasons the oxygen content of the right ventricular blood in the presence of an interventricular septal defect may be significantly higher than that of right auricular blood In the patent ductus arteriosus the pulmonary artery blood oxygen is significantly higher than that of the right ventricle owing to the shunt of arterial blood from the aorta The pulmonary artery blood flow may be determined from the oxygen content of pulmonary arterial and pulmonary venous blood and for full details the reader is referred to the work of Dexter *et al* (1947) Bing (1947) has calculated the collateral blood supply through bronchial arteries in cyanotic cases by a combination of catheter and respiratory methods Depending

upon the abnormality present there may exist considerable variation in pulmonary arterial blood With large interventricular septal defects or with the patent ductus mixture may be incomplete and multiple sampling from a pulmonary artery branch may be necessary to yield an average figure The oxygen content of pulmonary venous blood is calculated from a sample obtained from a distal branch of the pulmonary artery the furthermost point that the tip of the catheter will reach

The results obtained by these methods of examination have been admirably tabulated by Bing Handelsman and Campbell (1948) on the appended table

RESULTS OF CATHETERIZATION IN CONGENITAL HEART DISEASE

	Pressures Mm of Mercury	*Analysis of Gases in Blood*
Normal	RA 5 0 RV–25 0 PA–25 8	RA–RV–PA within 0 5 vol per cent
Tetralogy of Fallot Preoperative	RV elevated PA lower than RV	O in RV significantly higher than in RA
Postoperative	RV elevated PA lower than RV	O_2 in RV significantly higher than in RA
Eisenmenger s Complex	RV elevated PA systolic and diastolic elevated	O_2 in RV markedly higher than in RA
Pulmonary Stenosis	RV elevated PA lower than RV	RA–RV
Single ventricle with pulmonary stenosis	RA and RV elevated	O_2 in RV markedly higher than in RA (greatest difference observed)
Tricuspid Stenosis	RA elevated	O in RA significantly higher than in SVC
Patent ductus Arteriosus	RV PA normal	O_2 in PA significantly higher than in RV
I olated septal defects	Normal or elevated	RA significantly exceed SVC or RV exceeds RA

RA right auricle RV right ventricle PA pulmonary artery PC pulmonary capillary S systemic circulation SVC superior vena cava

CHAPTER XXI

DIFFERENTIAL DIAGNOSIS

The Recognition of Congenital Heart Disease

Congenital defects of the heart and great vessels are not uncommon Many of the abnormalities would be unimportant were it not for their liability to involvement in a bacterial endocarditis Until quite recent times a diagnosis of congenital heart disease was considered to be adequate and it was exceptional to attempt a precise anatomical diagnosis The recent developments in surgical treatment of some anomalies and the prospect that careful study of cases may furnish additional advances in treatment makes it important that diagnosis should be as precise as possible

The recognition of congenital heart disease usually offers no great difficulty and it should be possible to assign a case to its particular group and in many cases arrive at the diagnosis of the anatomical lesion present Whilst exact diagnosis is possible in a number of anomalies on purely clinical grounds corroborative and in some cases diagnostic or even pathognomonic evidence is furnished by the radio graph and electrocardiogram

In the general investigation of a case of congenital heart disease a careful history is essential Special reference should be made to symptoms and the order of their appearance and as to the age when a lesion was first suspected or diagnosed

A history of a heart lesion since infancy affords very valuable evidence In this connection in the British Isles the routine school entrance record is a valuable source of information In some cases some intercurrent infectious disease such as measles has led to the discovery of a heart lesion A definite heart lesion encountered before the age of 5 is almost presumptive evidence of its congenital etiology The maximum incidence of the rheumatic infection is between the ages of 6 and 12 consequently there should be a careful inquiry as to rheumatic antecedents Further as most congenital heart lesions arise before the eighth week of intra uterine life the general question of the mother s health during the early weeks of gestation should be explored if only for academic interest If this were done as routine and data collected concerning the time of occurrence duration and nature of infections or illness during the period of pregnancy much light might be thrown upon the problem of foetal endocarditis and upon the genesis of cardiac defects In practice it is rare to obtain a history

of maternal disease and above all of rubella *It is interesting to note* the position of the child in the family and the age of its parents at the time of its birth

Points that may be elicited in the early history of the case refer to *whether the child was born a* blue baby or as to the occurrence of periods of cyanosis especially in relation to feeding or to respiratory infections It may here be emphasized that epilepsy is quite common in congenital heart disease and conversely epileptic fits frequently masquerade as heart attacks the coincidence of a systolic murmur and the cyanosis of the convulsive phase leading to an error in diag nosis Specific questions as to the occurrence of convulsive attacks *should always be asked The presence of either established or episodal* cyanosis in the young child is usually due to congenital heart disease

The presence of other congenital abnormalities elsewhere is of importance and may give a clue as to the etiology of a particular case This association may perhaps have been over emphasized by some authors The widely divergent figures of incidence are mentioned elsewhere Nevertheless it is always advisable to search for these associated abnormalities The abnormalities most often associated with a congenital heart lesion are deformities of the chest wall in particular Harrison s sulcus accessory nipples mongolism and arachnodactyly In both of these latter conditions the commonest *heart lesion is a defect of the interauricular septum and consequently* heart signs may be minimal or possibly late in obvious appearance Again the signs of congenital heart disease may be masked by rheu matic heart disease which is prone to accompany certain defects

The proper appreciation of physical signs is of fundamental import ance Many cases in the acyanotic group remain undiagnosed during life or only recognized at a late age because symptoms are few and the patient has adapted himself to his condition Some cases are assumed to be rheumatic owing to over emphasis of the teaching of carditis without other rheumatic manifestations Because of marked and noisy physical signs a quite unnecessary gravity may be attributed *to cases with minor anomalies* The conception still lingers that the congenital cardiac case is inevitably a puny child with extreme cyanosis Therefore in examining a case it is essential to note the murmurs present their distribution and propagation and their sites of maximum intensity *A thrill must always be sought in all positions of the patient* and in inspiration and expiration As a part of the routine examination in all cases it is wise to feel the femoral arteries and note if pulsation is present Murmurs and thrills in abnormal positions which do not correspond to those found in acquired valvular disease are charac teristic The physical signs in congenital heart disease particularly in the acyanotic group may be peculiarly well marked and have a remark *able constancy* over the years although there may occasionally be

gradual regression and ultimate disappearance of the physical signs of certain abnormalities Attention should also be paid to the aortic and pulmonary second sounds as they may be important when exact diagnosis is being considered Finally a heart has not been completely examined until the blood pressure has been taken and the presence of hypertension or a very low pressure may be of diagnostic significance The occurrence of infective endocarditis may suggest the possible presence of a congenital cardiac anomaly if there is no history of antecedent rheumatism

There remains the laboratory methods of diagnosis the X ray and the electrocardiogram The use of these methods is invaluable and may enable a complete diagnosis to be made The presence of an abnormal cardiac silhouette and in particular abnormalities an undue prominence of the pulmonary arc are of suggestive importance These aspects of examination are fully discussed under the individual lesions The electrocardiogram which is usually not a specific diagnostic procedure serves to show right or left ventricular preponderance and abnormalities of conduction and so affords important confirmatory evidence It may be necessary to study the case further with circulation rate cardiac catheterization and contrast radiography but such examinations are of exceptional employment

The Significance of a Systolic Murmur Recognition of the presence or absence of heart disease is of very great importance to the patient Diagnosis is largely based upon the presence of murmurs in certain situations and with certain characteristics The factors in the production of a murmur are numerous the most important being the passage of blood from an area of small diameter into an area of greater diameter This results in the formation of jet eddies which are the most audible if they impinge upon the walls of the heart or great vessels Similarly abnormal structures such as chordae or vegetations may vibrate in the stream of blood and give rise to murmurs The loudness of the murmur depends also upon the velocity of the blood stream passing through a constriction this being determined by the difference *of pressure on the two sides of the constriction If for any reason the* velocity of the blood stream is increased as by exercise the murmur may be intensified Where there is a very small difference in pressure between the two sides the velocity of the blood stream is diminished and a murmur may be absent or if present it may under the influence of stresses tending towards the equalization of pressures between the two sides become diminished or disappear Thus in the interauricular septal defect the pressure relations between the two auricles are such that a murmur is generally absent despite the presence of a shunt Changes in the shape of a valve or obstruction may also modify the character of the murmur present and funnel shaped valvular orifices are especially important in this respect In the funnel shaped deformity

so often found in aortic stenosis the funnel in this case directed with the stream of blood the murmur may be loud in systole A regurgitant murmur which post mortem examination suggests might have been present is often faint or inaudible and is damped by the funnel now opposed to the regurgitant stream and perhaps similarly affected by the funnel shaped aortic conus likewise disposed Such instances might be multiplied as in the case of pulmonary stenosis where actual recognition of pulmonary insufficiency is relatively rare

The term functional murmur is applied to those murmurs which are produced in the absence of deformity of the valves or septa or indeed of any part of the heart It may refer to murmurs which are exocardial and which may be produced by friction between the pleura and pericardium A cardiorespiratory murmur which characteristically varies with the phase of respiration and is perhaps abolished on deep inspiration is probably caused by the movement of air within the alveoli that part of the lung in contact with the heart Many murmurs heard about the base of the heart may be explained on the above grounds In addition it seems conceivable as Thayer (1925) pointed out that the passage of blood through the relatively rigid valvular rings into the more distensible vessels beyond may reasonably be a cause of murmur formation Relative mitral stenosis has been discussed by Bramwell Observation shows the extreme frequency of murmurs in the traditional valvular sites Indubitably a systolic murmur may appear in childhood in association with anaemia or with infections and being due to relative insufficiency the result of stretching of the valve rings may properly be termed functional murmurs

Systolic murmurs are quite commonly present in infancy apart from recognizable pathology Lyon *et al* (1940) found a murmur in 1 9 per cent of newborn infants Of 147 cases with a systolic bruit ninety two of which were followed into later life four died and at post mortem on two cases there was congenital heart disease Fourteen had persistent systolic bruits thought to be some form of congenital heart disease and of the remaining seventy four three cases had functional bruits or extrasystoles Cox (1948) found that 5 per cent of 630 children had a systolic bruit at some time The murmur was most often recognized when the child had attained the age of nine months or more that is to say at the time of transition from the vegetative to the more active state It seems clear that some of these were cases of congenital heart disease but they were not differentiated It is quite certain that a patent ductus or an interauricular septal defect may not be the cause of murmurs in the neo natal period On the other hand grave cardiac anomalies are early recognized because they give rise to both symptoms and signs

In regard to later childhood Holt (1932) states that a systolic murmur is present in about 50 per cent of normal children Other authorities

do not place the incidence so highly but are of the opinion that a systolic murmur is frequent in otherwise normal children The actual incidence of organic heart disease in childhood to judge from the reports of school medical officers is a trifle over 1 per cent There is thus a wide gap between the cases presenting signs which might be attributed to organic disease and the case with recognizable cardiac damage This serves to emphasize the importance of a close consideration and observation of cases presenting murmurs Of recent years there has been a swing of the pendulum towards minimizing the significance of a systolic murmur although actual statistics show a greatly increased mortality in the group with such murmurs The generally accepted criteria of organic heart disease are enlargement of the heart with constant physical signs a diastolic murmur being always indicative of cardiac damage A diastolic murmur may be a transient event during the course of the rheumatic infection but usually in childhood it signifies mitral or aortic disease The types of murmur giving the greatest difficulty in childhood are the functional and cardiorespiratory murmurs These are as important to the child as an organic murmur because their wrongful interpretation leads to much unnecessary restriction and may induce invalidism or an inferiority complex during the formative stages of a child s career Consequently all the criteria which may be employed in judging such a murmur should be applied to these cases Inconstancy and variation with position and phase of respiration are important in this respect Familiarity with the cardiac auscultatory signs in childhood soon removes most of the difficulty in the interpretation of such a murmur Recent advances in phonocardiography hold promise of being of the greatest help in this respect

Undoubtedly in certain cases of congenital heart disease the physical signs may undergo considerable regression if observation is extended over a period of years It is our experience that a thrill may disappear or be inconstant or a murmur become so altered as to become indistinguishable from a functional murmur Sometimes the murmur may entirely disappear as has been discussed under the maladie de Roger The regression of physical signs has been statistically examined by Wilson (1938) who finds that in a group of congenital cardiac cases observed over a long period of 30 per cent the murmur changed in character and transmission and in seven instances became indistinguishable from the cardiorespiratory murmur Even cyanosis disappeared in some of the cases Wilson however emphasized that the cardiac silhouette as observed fluoroscopically remains unchanged Wilson (1934) insists that a teleradiogram in the frontal plane is inadequate for the diagnosis of moderate degrees of enlargement and that only with the oblique view in addition can an accurate determination of the size of the heart be made

CLINICAL DIAGNOSIS OF INDIVIDUAL LESIONS A summary is given below of the principal diagnostic points in certain lesions that are recognizable Reference should also be made to the classification of Abbott where defects are arranged according to the presence or absence of cyanosis and the degree of cyanosis presented

SUBAORTIC AND AORTIC STENOSIS A systolic thrill is felt in the aortic area and a systolic murmur is conducted along the vessels into the neck and axillae Thrill and murmur are maximum in the second right space In aortic stenosis there may be alterations in the aortic second sound There is a small radial pulse in marked contrast to a rather forcible apex beat The blood pressure is low Under development is most likely to occur in aortic stenosis and may amount to actual dwarfism The radiograph shows a slightly enlarged left ventricle and the electrocardiogram normal or left axis deviation

COARCTATION OF THE AORTA A systolic murmur is present over the base of the heart and is characteristically heard along the borders of the vertebral column It may also be heard over the visible and palpable vessels forming the collateral circulation Pulsation in the abdominal aorta and femoral vessels is reduced or absent Hypertension in the arms and low blood pressure in the legs is the rule The radiograph may show the pathognomonic notching of the under surface of the ribs and there is usually a small aortic knuckle Left axis deviation may be present but it is more common to find a normal axis

ISOLATED VENTRICULAR SEPTAL DEFECTS A systolic murmur occupies the whole of systole and is usually loud and rasping in marked contrast to the absence of symptoms The murmur is loudest in the fourth left space close to the sternum and is heard in diminished intensity at the periphery A thrill frequently accompanies the murmur There is no definite radiological picture The electrocardiogram is normal but rare cases may show conduction disturbances such as bundle branch block or complete heart block This type of congenital defect may give the greatest difficulty in diagnosis particularly in those cases where a thrill is absent Recent developments in phonocardiography hold promise of being of great value in the diagnosis of these cases

PATENT DUCTUS ARTERIOSUS In its most typical form a machinery or continuous murmur is heard in the second left space close to the sternum and is conducted towards the left clavicle The pulmonary second sound is accentuated or reduplicated Evidence of dilatation of the pulmonary artery is furnished by Gerhardt s ribbon dullness in the second and third left interspaces There may be a characteristic radiograph of a dilated pulmonary artery but this is not inevitable Hilar pulsation may be prominent The electrocardiogram shows no abnormality

PERSISTENT RIGHT AORTIC ARCH The diagnosis is mainly radiological but there may be dullness to the right of the sternum in the

upper intercostal spaces The radiograph gives the pathognomonic picture of displacement of the oesophagus and trachea to the left with absence of the aortic knob in its usual situation

AURICULAR SEPTAL DEFECTS Dyspnoea is habitual Pallor with slight cyanosis may be present the cyanosis being variable and more pronounced with exertion Symptoms characteristically appear in the second and third decades *The heart is enlarged There are no typical* thrills or murmurs but both may be present or absent A diastolic murmur of pulmonary incompetence may be present Occasionally the murmur of mitral stenosis may be recognized but not so frequently as post mortem experience would anticipate Alone in congenital heart lesions auricular fibrillation is not infrequent and it occurs in those cases complicated by mitral stenosis or hypertension The radiological picture shows gross cardiac enlargement with a large prominent pulmonary arc large comma right pulmonary branch and a small aortic knuckle Right axis is present in the electrocardiogram

PULMONARY STENOSIS WITH A CLOSED INTERVENTRICULAR SEPTUM Cyanosis may be absent until the third decade but once installed is usually progressive There is a systolic murmur and thrill in the second left space and the pulmonary second sound may be diminished or absent The radiograph shows a prominent pulmonary arc and hyper trophy of the right ventricle The electrocardiogram always shows right axis deviation

THE TETRALOGY OF FALLOT Cyanosis appears early is progressive and becomes extreme Symptoms are mainly dependent upon the cyanosis A systolic murmur may be present in the second left space but rarely of the intensity shown in isolated pulmonary stenosis The radiograph shows a coeur en sabot silhouette with a concavity at the site of the pulmonary arc corresponding to the hypoplasic pulmonary artery The hilar vessels are small and there is absence of congestion in the lung fields The electrocardiogram exhibits a marked degree of right axis deviation

THE EISENMENGER COMPLEX Cyanosis appears late and is rarely marked A systolic murmur may be present and a diastolic murmur of pulmonary incompetence has figured in the reported cases The diag nosis is mainly radiological and is based upon enlargement of the pulmonary artery Right axis is present in the electrocardiogram

CHAPTER XXII

GENERAL PROGNOSIS AND TREATMENT

The problems of prognosis are so varied that some consideration has been given to them in the discussion of the individual defects There are however certain general principles which may be applied to the different groups of cases The congenital heart defect differs from all other heart lesions in that it is permanent and stable from birth The patient becomes adapted to his lesion and is able to tolerate much that would be virtually impossible had the defect been acquired at a later age The relation of the defect to the heart as a whole may vary with the growth of the heart and a defect of significance in infancy may become relatively unimportant as the heart and the organism generally matures The converse may be true as witness the liability of the subject of coarctation to break down in the third decade at a time when activity is greatest A valvular stenotic lesion may become a severe handicap with growth and increased activity It may even be inhibitory to growth as in the dwarfism of aortic stenosis or in the slender gracile bodily conformation and retarded development of the patent ductus arteriosus with a large shunt The prognosis thus turns upon the stresses and strains to which the anomalous structures may be subject and the degree of interference to the circulation imposed by the lesion These stresses are most likely to occur in infancy or during early adult life

Factual knowledge of the prognosis of congenital heart disease is largely based upon post mortem statistics These are open to the criticism that in general only remarkable or peculiar cases are reported and no account is taken of the living The incidence of congenital heart disease is to some extent influenced by the interest of clinician and pathologist and is naturally higher in those hospitals devoted to infants and children Lastly some of the well known series such as that of Abbott (1931) comprise a relatively high number of neonatal deaths some at an early age when an abnormality such as a patent ductus arteriosus can hardly be regarded as an anomaly

The ever present danger in all groups is infective endocarditis The risks appear to be greater in the acyanotic group and Abbott s statistics show an incidence of 22 per cent cases with infective endocarditis in this group compared with 14 per cent in the cyanotic groups The disparity in incidence between the two groups may perhaps only be superficial for as Perry (1937) points out the acyanotic case lives longer than the cyanotic and consequently the risk is greater On the

other hand the acyanotic group furnish more obstructive lesions and sites of vigorous shunts Gelfman and Levine (1942) in their analysis of patients over two years of age placed significant defects according to risk as follows interauricular septal defects none interventricular septal defects 57 per cent patent ductus arteriosus 20 per cent bicuspid valves 17 4 per cent tetralogy of Fallot 29 per cent pulmonary stenosis 29 per cent The source of infection is generally the mouth and upper air passages Other potential sources of infection are mainly surgical and in relation to appendix abscess and post operative femoral thrombosis Infection from these sources may not only give rise to infective endocarditis but also to cerebral abscess in the cyanotic case with a dextroposed aorta

The incidence of pulmonary tuberculosis in congenital heart disease has long been a subject of discussion and controversial opinion Rokitansky (1875) stated that pulmonary tuberculosis and heart disease were incompatible especially when the latter was associated with cyanosis This contention can hardly be maintained at the present time Peacock (1866) mentions nine cyanotic cases with pulmonary tuberculosis The recent French literature furnishes a number of examples *Abbott (1931) lists 110 cases with pulmonary stenosis* of whom 22 per cent had phthisis Auerbach and Stemmerman (1944) have studied a group of thirteen cases of whom ten had pulmonary stenosis (in five a component of the tetralogy) and three acyanotic congenital heart disease all with phthisis In the acyanotic group Abbott s figures disclose 5 5 per cent with phthisis the highest sectional incidence being in the interventricular septal defect where of sixty two cases 14 5 per cent died with pulmonary tuberculosis Gloyne (1936) found seven cases of phthisis in twenty one cases of congenital heart disease and thought that it was uncommon in the cyanotic case In our own experience tuberculosis and cyanotic congenital heart disease has occurred ten times and six times in acyanotic disease It seems therefore that there is some increased liability for the cyanotic case with pulmonary stenosis to develop phthisis This incidence requires explanation for it has been argued that cyanosis should protect the subject from pulmonary infection as exemplified by the low incidence of phthisis in mitral stenosis In this latter condition it has been suggested that an excess of blood in the pulmonary circulation may be a factor in immunity because pulmonary congestion by inducing pulmonary fibrosis might well protect the subject against the tubercular infection Such considerations are hardly applicable to the usual cyanotic case where cyanosis depends upon the quite different factors of a shunt and above all upon inadequate blood supply to the lungs Modern studies of the circulation in the lungs have shown that blood circulates in them at a relatively low pressure and the position of the subject largely determines the amount of blood that reaches the apex

or base In recumbency when the effects of gravity are removed the circulation in the lungs increases venous pressure rises and the output of the heart may be increased by as much as 30 per cent Lymph flow increases with rise of venous pressure and when the patient is recumbent removal of bacteria from the lungs is effective at both the apices and bases In the upright or orthopnoeic position venous pressure is low and arterial flow reduced at the apices of the lungs The origin of the right pulmonary artery at an acute angle to the direction of flow in the pulmonary conus and its tortuous path round the aorta leads to a definite fall in pressure along its course and Dock (1947) states that the apical third of the right lung and the apical fourth of the left lung have practically no effective flow for two thirds of the day in sedentary people Lymph flow also decreases and accordingly antibodies formed in the body and antibiotics given parenterally do not reach these areas An ideal situation is therefore created for the development of tuberculosis in these areas where dust bacteria and toxins cannot easily be removed It seems therefore that pulmonary arterial inflow is the crucial factor for pulmonary stenosis lowers pulmonary arterial pressure whereas in mitral stenosis pulmonary arterial pressure is considerably elevated Other changes such as oxygen unsaturation and increased carbon dioxide tension or alterations in the Ph of the blood have been discussed by Gloyne and Shiskin (1937) In our own opinion tuberculosis in the cyanotic case is more common than has been hitherto supposed The general clinical features and course of pulmonary tuberculosis in congenital heart disease are no different from those in cases without heart disease The patient has a good prospect of early recognition of his disease because of the frequency with which he undergoes X ray examination on account of his heart abnormality For treatment bed rest may be sufficient but the presence of a heart lesion is not a contradiction to collapse therapy This has successfully been carried out on several occasions It is well to institute collapse therapy at an early stage before adhesions have formed The presence of adhesions may have a certain value in providing a collateral circulation with the tetralogy of Fallot and the actual production of adhesions has a place in the treatment of this condition The newer Monaldi methods of closing cavities may have considerable value in the treatment of these patients as they involve little lessening of vital capacity

The incidence of the rheumatic infection in cases of congenital heart disease is a matter of interest because acquired rheumatic heart disease appears to be fairly common in congenital heart cases that reach adult life This is particularly true of the interauricular septal defect (Roesler 1934 Bedford Papp and Parkin 1941 Burrett and White 1945) So high is the incidence of mitral stenosis in the anomaly that it is justifiable to suspect that in a proportion of cases mitral stenosis may be

the primary event leading to the transformation of a potentially patent foramen ovale into a wide aperture Gelfman and Levine s series of congenital heart disease showed a superadded rheumatic infection in 14 per cent of cases A predilection to rheumatism has also been noted in respect to bicuspid aortic valves

With certain exceptions in those cases where there is no communication between the two sides of the heart and in the group with cyanose tardive the outlook is generally good and the subject should be encouraged to live as normal a life as possible Generally speaking the unfavourable signs are the presence of symptoms and of enlargement of the heart A diastolic murmur should always be considered unfavourable The activities allowed should be proportional to the subject s capabilities Where there is no communication between the two sides as in aortic stenosis or coarctation there is a risk of strain Coarctation demands especial consideration In many cases it is as well tolerated as other defects but certain danger periods are undoubtedly present These occur during early adult life when failure may supervene in response to strain Infective endocarditis claims a large number of cases There is liability to accidents due to hypertension such as rupture of the aorta or a cerebral catastrophe may result from the rupture of an associated cerebral aneurysm The coincidence of aortic incompetence is definitely unfavourable Severe hypertension signs of imminent failure and cardiac enlargement are indications for operation in the young The adult case should in the light of present knowledge and experience be judged on its merits In subaortic stenosis a lesion that offers very little impediment to the circulation the outlook is reasonably good although sudden death may occur here as in other forms of aortic stenosis It may be said of the acyanotic group as a whole that the prognosis is reasonably good when certain features are eliminated Unfavourable circumstances are the presence of gross enlargement of the heart this being most likely to be found in cases complicated by aortic incompetence or by high grade obstructive valvular lesions There is a considerable incidence of infective endocarditis The presence of high blood pressure and its complications must inevitably lead to some restriction Death in the group is most likely to be due to a complication or intercurrent disease Sudden death may occur in stenotic lesions and congestive failure is rare In the group with cyanose tardive the prognosis is quite good In the maladie de Roger statistics show that there is considerable risk of infective endocarditis Congenital heart block may be a complication but generally causes little inconvenience to the patient Isolated interventricular septal defects are usually entirely benign during the school period and no restrictions are necessary The patent ductus arteriosus demands more care In this lesion there is a considerable strain on the heart and further there is a high incidence of infective endocarditis Surgical

treatment may be confidently recommended in childhood and is urgent at any age if infection supervenes Little is known about the remote prognosis of the untreated ductus save that it is rare in the consulting room in adult life The matter is more fully discussed in Chapter IX

The auricular septal defect presents certain points of difference from the other types of congenital heart disease It may be entirely latent until the second or third decade and compatible with normal activity After that time cyanosis and failure tend to appear With the frequent incidence of rheumatic valvular disease and in particular mitral stenosis *the heart becomes grossly enlarged and auricular fibrillation* may occur Infective endocarditis rarely if ever occurs with the uncomplicated auricular septal defect It thus appears that each case of the cyanose tardive must be considered on its own merits and restrictions will occasionally be necessary Unfavourable circum stances are *the presence of symptoms attributable to the lesion* and the presence of enlargement of the heart Intercurrent infection may claim a large proportion of the members of this group and with the exception of the auricular septal defect the occurrence of congestive failure is relatively rare

In the cyanotic group the main factors for consideration are the degree of cyanosis and the severity of symptoms Cyanosis appearing in early childhood is of serious prognostic import although some regression of cyanosis may occur (Wilson 1936) Perry (1937) suggests that in the main cases showing cyanosis at or soon after birth rarely survive five years Cyanosis becoming permanent after five years usually results in death before puberty whilst those developing cyanosis after puberty may lead a comparatively long if restricted life There are however exceptions to this broad prognostic classification but in *general the period of active growth at puberty is a critical time* Observation of these cases over a period of years shows that the degree of cyanosis may remain singularly constant and suggests that there is a need for revision of the current ideas of prognosis in this group An isolated blood count has not the same significance as blood counts taken at intervals A rising blood count is of serious significance The lesions of the group that are best tolerated are the tetralogy of Fallot the Eisenmenger complex and a complete defect of the interventricular septum Infective endocarditis does not appear to be as frequent as in the acyanotic groups A possible reason for this is that survival is longer in the acyanotic group and there is consequently more oppor tunity to acquire a bacterial infection Each case must be considered on its merits bearing in mind the high mortality in early childhood from the severity of the defect The presence of symptoms is almost invariable in the group and the more severe these are the more serious the outlook Surgical treatment offers the only possible help for the patient and should be employed whenever it is possible to establish

that the fundamental physiological difficulty is an inadequate blood supply to the lungs

PREGNANCY Congenital heart disease is a comparatively rare finding in pregnant women because the majority of patients with serious congenital heart disease die before the age of puberty There are many isolated examples of congenital heart disease and pregnancy in the literature but few collected series Mendelson and Pardee (1941) have reported twenty of their own cases There was no mortality in their series contrasting with the 11 per cent mortality of 135 cases collected from the literature With the possible exception of coarctation preg nancy does not appear to be contra indicated in acyanotic congenital heart disease Naturally each case must be judged on its merits and unfavourable points are important cardiac enlargement signs of failure or of functional incapacity In our own experience we have observed uncomplicated pregnancy in a case of subaortic stenosis five cases of patent ductus arteriosus and three cases with an auricular septal defect There is a very definite risk in cases of coarctation of the aorta and Mendelson (1940) after the study of three cases of his own and twenty nine from the literature decided that if pregnancy was discovered early therapeutic abortion was desirable If termination was not feasible delivery should be effected by Caesarean section and the patient sterilized He observed that there was definite deterioration of the patient s condition during pregnancy and his analysis shows five deaths due to aortic rupture cerebral accidents cardiac failure or endocarditis Personal experience with four cases confirms the view that coarctation is a dangerous complication of pregnancy and preg nancy is best terminated by Caesarean section and sterilization per formed Each of these four cases were referred from antenatal clinics with a diagnosis of hypertension and the average parity was 2 5 The frequency of pregnancy in the Lutembacher syndrome is a feature of the reported cases of this group In cases such as the patent ductus interventricular or auricular septal defect there is a danger of reversal of the shunt if much haemorrhage occurs and these dangers should be watched for and forestalled or promptly treated by appropriate transfusion and other means

It is rare for pregnancy to be observed in the severe cyanotic case partly because few cases attain the reproductive age and partly on account of restrictions placed upon pregnancy in this state Shapiro and Simons (1934) were of the opinion that congenital heart disease even if associated with severe cyanosis was not in itself a contra indication to pregnancy Naturally the cyanotic case represents the most severe type of congenital heart anomaly and pregnancy is best terminated early If some compelling reason exists for continuing the pregnancy delivery should be by Caesarean section Experience in the surgery of cyanotic congenital heart disease shows that an anaesthetic

is well taken in these cases Uncomplicated pulmonary stenosis is well tolerated and requires no special precautions

The management of pregnancy should follow the usual lines and depends largely upon the functional capacity of the patient The patient must be carefully observed and rest and digitalis used as necessary together with any obstetrical procedures likely to minimize strain during the course of pregnancy and labour Symptoms may appear or be aggravated from the fourth month onwards The presence of failure should not necessarily be regarded as an indication for Caesarean section or for early induction of labour Neither should the presence of a heart lesion be considered as an absolute indication for operative treatment Digitalis should be used if failure appears and the patient kept in bed Rest should be encouraged in the last two months of pregnancy

LIFE ASSURANCE Sometimes an acyanotic case with marked physical signs and absence of symptoms may present himself for life assurance Obviously certain types of congenital heart disease such as the cyanotic case or coarctation of the aorta are unacceptable A well tolerated patent ventricular septum or even a patent ductus arteriosus may be accepted for a limited term of years not exceeding the age of 40 (East 1937) A heavy load is essential to cover the risk of an infective endocarditis

MEDICO-LEGAL Rarely cases of congenital heart disease may have a medico legal importance A large number of cases of congenital heart disease come under the observation of the physician and are warned as to the amount and nature of work that they may undertake being usually forbidden to engage in strenuous work Cases of the acyanotic type and in particular cases of coarctation and aortic stenosis may become matters of litigation where symptoms have been brought on by effort made during working hours Where a fatal attack occurs presumed to be the result of some strenuous effort or where incapacity arises from similar causes compensation may be sought for illness or death arising out of or during the course of employment Infective endocarditis consequent upon infection of a small abrasion or punctured wound sustained during the course of employment may be compensatable A congenital hypoplasia of the aorta and of the coronary arteries has been the subject of a claim against a school which was held to be liable for the sudden death of a pupil Apart from the above aspects sudden death in congenital heart disease may be a matter of interest to the coroner T Skene Keith (personal communication) in 1 280 autopsies performed for the coroner found twenty two (1 7 per cent) cases of congenital anomaly seventeen of these cases being below the age of one year

TREATMENT The very nature of a congenital cardiac defect consisting of a stable anatomical change in the heart makes treatment a matter

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This Book

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By HUGH JOLLY M A , M D (CAMB) M R C P

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